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09/472688  
12/27/99

**IN THE UNITED STATES PATENT AND TRADEMARK OFFICE**

FIRST-NAMED INVENTOR: Shimkets  
FOR: NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE  
POLYMORPHISMS AND METHODS OF USE THEREOF

December 27, 1999  
Boston, Massachusetts

Box PATENT APPLICATION  
Assistant Commissioner for Patents  
Washington, D.C. 20231

**REQUEST FOR FILING A CONTINUING UTILITY APPLICATION  
UNDER 37 C.F.R. §1.53(b)**

1. This is a request for filing a continuing application under 37 C.F.R. §1.53(b).  
(Check the appropriate boxes and supply the requisite information)  
This application is a:  
☐ Continuation; ☐ Divisional; ☒ Continuation-in-part of prior application U.S.S.N. 09/442,849.
2. Priority to the above application(s) is claimed under:  
☒ 35 U.S.C. §120  
Prior application information:  
Examiner: Not Yet Assigned; Group Art Unit: Not Yet Assigned.  
☐ 35 U.S.C. §119  
Priority of application Serial No. \_\_\_\_\_, filed on \_\_\_\_\_ in \_\_\_\_\_ is claimed under  
35 U.S.C. §119.  
☐ The certified copy has been filed in prior application U.S.S.N. \_\_\_\_/\_\_\_\_, \_\_\_\_ on [Date].  
☐ The certified copy will follow.

**APPLICATION ELEMENTS:**

3. ☒ Specification and Drawings (Total pages: 265):  
Specification (251 pages); Claims (12 pages); Abstract (1 page); and  
Drawings: 1 sheet; FIGS. 1.  
☐ Formal  
☒ Informal
4. ☒ Nucleotide and/or Amino Acid Sequence Submission:  
☒ Computer-readable copy  
☒ Paper copy (identical to computer-readable copy)  
☒ Statement verifying paper copy identical to computer-readable copy
5. ☐ Microfiche Computer Program (*Appendix*)
6. ☒ Oath or Declaration (Total pages: 3):  
(a) ☒ Unsigned (original)

**FIRST-NAMED INVENTOR:** Shimkets  
**Request for Continuing Nonprovisional Application (37 C.F.R. §1.53(b))**

(b) ☐ Copy from a prior application (37 C.F.R. §1.63(d))

7. ☐ Incorporation by Reference (*can use if Box 6(b) is checked*)

The entire Disclosure of the prior application, from which a copy of the oath or declaration is supplied under Box 6(b), is considered as being part of the disclosure of the accompanying application and is hereby incorporated by reference therein.

**ACCOMPANYING APPLICATION PARTS:**

8. ☐ English Translation Document (*if applicable*)

9. ☐ Information Disclosure Statement (IDS)

☐ Copy of IDS and PTO-1449 (\_\_\_ pages)

☐ Copies of references cited

10. ☐ Statement Claiming Small Entity Status

☐ Copy of Statement filed in prior application (Status still proper and desired)

11. Fee Calculation:

☐ A Preliminary Amendment is enclosed herewith. Please enter the claim amendments prior to calculating the filing fee.

CLAIMS AS FILED					
Claims	Number Filed	Basic Fee Allowance	Number Extra	Rate	Basic Fee 37 C.F.R. 1.16(a) \$760.00
Total Claims (37 C.F.R. 1.16(c))	44	- 20 =	24	\$ 18.00	\$432.00
Independent Claims (37 C.F.R. 1.16(b))	13	- 3 =	10	\$78.00	\$780.00
Multiple Dependent Claim(s), if any (37 C.F.R. 1.16(d))				\$260.00	0
				SUBTOTAL:	\$1972.00
				Reduction by 50% for filing by small entity:	- \$986.00
				<b>TOTAL FEE:</b>	\$986.00

12. ☒ A check in the amount of **\$986.00** is enclosed.

13. ☐ Deletion of Inventor(s)

This application is filed by fewer than all the inventors named in the prior application, 37 C.F.R. §1.53(d)(4).

☐ DELETE the following inventor(s) named in the prior nonprovisional application:

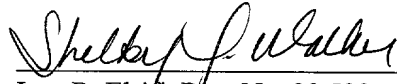
\_\_\_\_\_  
\_\_\_\_\_

☐ The inventor(s) to be deleted are set forth on a separate sheet attached hereto.

FIRST-NAMED INVENTOR: Shimkets  
Request for Continuing Nonprovisional Application (37 C.F.R. §1.53(b))

14. ☒ The Commissioner is hereby authorized to credit overpayments or charge the following fees to Deposit Account No. 50-0311, Ref. No. 15966-534C CIP1:
- ☒ Fees required under 37 C.F.R. §1.16;
  - ☒ Fees required under 37 C.F.R. §1.17;
  - ☒ Fees required under 37 C.F.R. §1.18.
15. ☒ Return Receipt Postcard Enclosed.
16. ☒ Other Documents Enclosed:
- ☐ Change of Attorney Address In Application.
  - ☒ Limited Recognition under 37 C.F.R. § 10.9(b) for Michel Morency.

Respectfully submitted,



Dated: December 27, 1999

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TRADOCS:1277638.1(RDTY01!.DOC)

## NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE POLYMORPHISMS AND METHODS OF USE THEREOF

10

### RELATED APPLICATIONS

This application is a continuation-in-part of U.S.S.N. 09/442,849, filed November 17, 1999, which claims priority to USSN 09/442,129 and USSN \_\_\_\_, both filed November 16, 1999, all of which are entitled "Nucleic Acids Containing Single  
15 Nucleotide Polymorphisms and Methods of Use Thereof" and naming Richard Shimkets and Martin Leach as inventors, and to USSN 60/109,024, filed November 17, 1998. The contents of these applications are incorporated herein by reference in their entirety.

### FIELD OF THE INVENTION

The invention relates generally to nucleic acids and polypeptides and in particular  
20 to the identification of human single nucleotide polymorphisms based on at least one gene product that was not previously described.

### BACKGROUND OF THE INVENTION

Sequence polymorphism-based analysis of nucleic acid is generally based on alterations in nucleic acid sequences between related individuals. This analysis has been  
25 widely used in a variety of genetic, diagnostic, and forensic applications. For example, polymorphism analyses are used in identity and paternity analysis, and in genetic mapping studies.

Several different types of polymorphisms in nucleic acid have been described. One such type of variation is a restriction fragment length polymorphism (RFLP). RFLPS  
30 can create or delete a recognition sequence for a restriction endonuclease in one nucleic



- 5 acid relative to a second nucleic acid. The result of the variation is in an alteration the relative length of restriction enzyme generated DNA fragments in the two nucleic acids.

Other polymorphisms take the form of short tandem repeats (STR) sequences, which are also referred to as variable numbers of tandem repeat (VNTR) sequences. STR sequences typically that include tandem repeats of 2, 3, or 4 nucleotide sequences that are  
10 present in a nucleic acid from one individual but absent from a second, related individual at the corresponding genomic location.

Other polymorphisms take the form of single nucleotide variations, termed single nucleotide polymorphisms (SNPs), between individuals. A SNP can, in some instances, be referred to as a “cSNP” to denote that the nucleotide sequence containing the SNP  
15 originates as a cDNA.

SNPs can arise in several ways. A single nucleotide polymorphism may arise due to a substitution of one nucleotide for another at the polymorphic site. Substitutions can be transitions or transversions. A transition is the replacement of one purine nucleotide by another purine nucleotide, or one pyrimidine by another pyrimidine. A transversion is  
20 the replacement of a purine by a pyrimidine, or the converse.

Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Thus, the polymorphic site is a site at which one allele bears a gap with respect to a single nucleotide in another allele. Some SNPs occur within, or near genes. One such class includes SNPs falling within  
25 regions of genes encoding for a polypeptide product. These SNPs may result in an alteration of the amino acid sequence of the polypeptide product and give rise to the expression of a defective or other variant protein. Such variant products can, in some cases result in a pathological condition, *e.g.*, genetic disease. Examples of genes in which a polymorphism within a coding sequence gives rise to genetic disease include sickle cell  
30 anemia and cystic fibrosis. Other SNPs do not result in alteration of the polypeptide product. Of course, SNPs can also occur in noncoding regions of genes.

SNPs tend to occur with great frequency and are spaced uniformly throughout the

- 5 genome. The frequency and uniformity of SNPs means that there is a greater probability that such a polymorphism will be found in close proximity to a genetic locus of interest.

#### SUMMARY OF THE INVENTION

The invention is based in part on the discovery of single nucleotide polymorphisms (SNPs) in regions of human DNA.

- 10 Accordingly, in one aspect, the invention provides nucleic acid sequences comprising nucleic acid segments of both publicly known and novel genes, including the polymorphic site. The segments can be DNA or RNA, and can be single- or double-stranded. Preferred segments include a biallelic polymorphic site.

- 15 The invention further provides allele-specific oligonucleotides that hybridize to a segment of a fragment shown in Table 1, column 4, or its complement. These oligonucleotides can be probes or primers. Also provided are isolated nucleic acids comprising a sequence shown in Table 1, column 4, in which the polymorphic site within the sequence is occupied by a base other than the reference bases shown in Table 1, columns 5 and 6.

- 20 The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites shown in Table 1. Optionally, a set of bases occupying a set of polymorphic sites shown in Table 1 is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype.

- 25 In another aspect, the invention provides an isolated polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, *e.g.*, a nucleotide sequence which includes one or more of the polymorphic sequences shown in Table 1 and which includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long as it includes the polymorphic site. The polynucleotide may  
30 alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of these sequences, or a fragment of the complementary nucleotide

- 5 sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

The polynucleotide can be, *e.g.*, DNA or RNA, and can be between about 10 and about 100 nucleotides, *e.g.* 10-90, 10-75, 10-51, 10-40, or 10-30, nucleotides in length.

- 10 In preferred embodiments, the polymorphic site in the polymorphic sequence includes a nucleotide other than the nucleotide listed in Table 1, column 5 for the polymorphic sequence, *e.g.*, the polymorphic site includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

- 15 In other embodiments, the complement of the polymorphic site includes a nucleotide other than the complement of the nucleotide listed in Table 1, column 5 for the complement of the polymorphic sequence, *e.g.*, the complement of the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

- 20 In some embodiments, the polymorphic sequence is associated with a polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

- 25 In another aspect, the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic sequences recited in Table 1, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence. Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic sequences in Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of

- 5 the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

In some embodiments, the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide. The second polynucleotide can be, *e.g.*, (a) a nucleotide sequence comprising one or more polymorphic sequences in Table 1, wherein  
10 the polymorphic sequence includes the nucleotide listed in Table 1, column 5 for the polymorphic sequence; (b) a nucleotide sequence that is a fragment of any of the polymorphic sequences; (c) a complementary nucleotide sequence including a sequence complementary to one or more polymorphic sequences disclosed herein in Table 1; and  
15 (d) a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

The oligonucleotide can be, *e.g.*, between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

The invention also provides a method of detecting a polymorphic site in a nucleic  
20 acid. The method includes contacting the nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected shown in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The  
25 method also includes determining whether the nucleic acid and the oligonucleotide hybridize. Hybridization of the oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphic site in the nucleic acid.

In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in  
30 Table 1, column 5 for the polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for the polymorphic sequence.

5           The oligonucleotide can be, *e.g.*, between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

          In some embodiments, the polymorphic sequence identified by the oligonucleotide is associated with a nucleic acid encoding polypeptide related to one of the protein families disclosed herein. the polymorphic sequence is associated with a  
10   polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

15           In a further aspect, the invention provides a method of determining the relatedness of a first and second nucleic acid. The method includes providing a first nucleic acid and a second nucleic acid and contacting the first nucleic acid and the second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide  
20   other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The method also includes determining whether the first nucleic acid and the second nucleic acid hybridize to the oligonucleotide, and comparing hybridization of the first and second nucleic acids to the oligonucleotide. Hybridization  
25   of first and second nucleic acids to the nucleic acid indicates the first and second subjects are related.

          In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or when the complement of the  
30   polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 column for the polymorphic sequence.

5           The oligonucleotide can be, *e.g.*, between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

          The method can be used in a variety of applications. For example, the first nucleic acid may be isolated from physical evidence gathered at a crime scene, and the  
10   second nucleic acid may be obtained is a person suspected of having committed the crime. Matching the two nucleic acids using the method can establishing whether the physical evidence originated from the person.

          In another example, the first sample may be from a human male suspected of being the father of a child and the second sample may be from a child. Establishing a  
15   match using the described method can establishing whether the male is the father of the child.

          In another aspect, the method includes determining if a sequence polymorphism is the present in a subject, such as a human. The method includes providing a nucleic acid from the subject and contacting the nucleic acid with an oligonucleotide that hybridizes  
20   to a polymorphic sequence disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. Hybridization between the nucleic acid and the oligonucleotide is then determined. Hybridization of  
25   the oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphism in said subject.

          In another aspect, the invention provides an isolated polypeptide comprising a polymorphic site at one or more amino acid residues, and wherein the protein is encoded by a polynucleotide including one of the polymorphic sequences in Table 1, or their  
30   complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

5           The polypeptide can be, *e.g.*, related to one of the protein families disclosed herein. For example, polypeptide can be related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

          In some embodiments, the polypeptide is translated in the same open reading  
10   frame as is a wild type protein whose amino acid sequence is identical to the amino acid sequence of the polymorphic protein except at the site of the polymorphism.

          In some embodiments, the polypeptide encoded by the polymorphic sequence, or its complement, includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence, or the complement includes the complement of the nucleotide listed in Table 1,  
15   column 6.

          The invention also provides an antibody that binds specifically to a polypeptide encoded by a polynucleotide comprising a nucleotide sequence encoded by a polynucleotide including one or more of the polymorphic sequences in Table 1, or its complement. The polymorphic sequence includes a nucleotide other than the nucleotide  
20   recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

          In some embodiments, the antibody binds specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

25           Preferably, the antibody does not bind specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for the polymorphic sequence.

          The invention further provides a method of detecting the presence of a polypeptide having one or more amino acid residue polymorphisms in a subject. The  
30   method includes providing a protein sample from the subject and contacting the sample with the above-described antibody under conditions that allow for the formation of

- 5 antibody-antigen complexes. The antibody-antigen complexes are then detected. The presence of the complexes indicates the presence of the polypeptide.

The invention also provides a method of treating a subject suffering from, at risk for, or suspected of, suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, *e.g.*, a human, non-human primate, cat, dog, rat, mouse, cow,  
10 pig, goat, or rabbit. The method includes providing a subject suffering from a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence shown in Table 1, or its complement, and treating the subject by administering to the subject an effective dose of a therapeutic agent. Aberrant expression can include qualitative alterations in expression of a gene, *e.g.*, expression of a gene encoding a  
15 polypeptide having an altered amino acid sequence with respect to its wild-type counterpart. Qualitatively different polypeptides can include, shorter, longer, or altered polypeptides relative to the amino acid sequence of the wild-type polypeptide. Aberrant expression can also include quantitative alterations in expression of a gene. Examples of quantitative alterations in gene expression include lower or higher levels of expression of  
20 the gene relative to its wild-type counterpart, or alterations in the temporal or tissue-specific expression pattern of a gene. Finally, aberrant expression may also include a combination of qualitative and quantitative alterations in gene expression.

The therapeutic agent can include, *e.g.*, second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide  
25 present in the wild type allele. In some embodiments, the second nucleic acid sequence comprises a polymorphic sequence which includes nucleotide listed in Table 1, column 5 for the polymorphic sequence.

Alternatively, the therapeutic agent can be a polypeptide encoded by a polynucleotide comprising polymorphic sequence shown in Table 1, or by a  
30 polynucleotide comprising a nucleotide sequence that is complementary to any one of the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.



5           The therapeutic agent may further include an antibody as herein described, or an oligonucleotide comprising a polymorphic sequence shown in Table 1, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence,

10           In another aspect, the invention provides an oligonucleotide array comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1; a nucleotide sequence that is a fragment of any of the nucleotide sequence, provided that the fragment includes  
15 a polymorphic site in the polymorphic sequence; a complementary nucleotide sequence comprising a sequence complementary to one or more of the polymorphic sequences; or a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

          In preferred embodiments, the array comprises 10; 100; 1,000; 10,000; 100,000 or  
20 more oligonucleotides.

          The invention also provides a kit comprising one or more of the herein-described nucleic acids. The kit can include, *e.g.*, polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, *e.g.*, a nucleotide sequence which includes one or more of the polymorphic sequences shown in Table 1, and which  
25 includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long as it includes the polymorphic site. The polynucleotide may alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of the sequences, or a fragment of the complementary nucleotide sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

30           Alternatively, or in addition, the kit can include the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1, provided that the polymorphic

5 sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence. Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic  
10 sequences shown in Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 6. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

15 **BRIEF DESCRIPTION OF THE DRAWING**

FIG. 1 illustrates an example of the way in which SNP sites were identified in the present invention.

Unless otherwise defined, all technical and scientific terms used herein have the same meaning as commonly understood by one of ordinary skill in the art to which this  
20 invention belongs. Although methods and materials similar or equivalent to those described herein can be used in the practice or testing of the present invention, suitable methods and materials are described below. All publications, patent applications, patents, and other references mentioned herein are incorporated by reference in their entirety. In the case of conflict, the present specification, including definitions, will  
25 control. In addition, the materials, methods, and examples are illustrative only and not intended to be limiting.

Other features and advantages of the invention will be apparent from the following detailed description and claims.

**DETAILED DESCRIPTION OF THE INVENTION**

30 The invention provides human SNPs in sequences which are transcribed, *i.e.*, are cSNPs. Many SNPs have been identified in genes related to polypeptides of known

5 function. If desired, SNPs associated with various polypeptides can be used together. For example, SNPs can be grouped according to whether they are derived from a nucleic acid encoding a polypeptide related to particular protein family or involved in a particular function. Similarly, SNPs can be grouped according to the functions played by their gene products. Such functions include, structural proteins, proteins from which associated with  
10 metabolic pathways fatty acid metabolism, glycolysis, intermediary metabolism, calcium metabolism, proteases, and amino acid metabolism, etc. Specifically, the present invention provides a large number of human cSNP's based on at least one gene product that has not been previously identified. In contrast, and as defined specifically in the following paragraph, the cSNP's involve nucleic acid sequences that are assembled from  
15 at least one known sequence.

The present invention describes 651 distinct polymorphic sites, which are summarized in Table 1. Raw traces underlying sequence data were drawn from public databases and from the proprietary database of the Assignee of the present invention. The sequences were obtained by calling the bases from these traces, and included  
20 assigning "Phred" quality scores for each called base. For each allelic set, at the polynucleotide level, four or more nucleotide sequences were identified having at least partial overlap with one another.

As illustrated in FIG. 1, these four or more sequences could be clustered and assembled to make a consensus contig that included an ORF. In this way, the inventors  
25 found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by a SNP at a particular polymorphic site. In order to be confirmed as a SNP site, the nucleotide change from the consensus sequence had to occur in at least two individual sequences, and had to have a "Phred" score of 23 or higher at the site of the presumed SNP. Furthermore, in a window of 5 bases on either side of the SNP, no  
30 more than 50% mismatching with the consensus sequence was allowed. In the assembly leading to each of the contigs defining the allelic set, the SNP alleles occur in polynucleotides found in public databases.

5           It was found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by an SNP at a particular polymorphic site. These associations were not previously known.

          At the level of translation of an ORF contained in the contigs, allelic sets were identified in which one allele defines a known polypeptide sequence that includes the  
10   polymorphic site and another polypeptide allele is not previously known. Then, various associations of alleles are possible. For example, it is possible that an allelic pair is defined in a noncoding region of the contig containing an ORF. In such cases the inventors believe that the invention resides in the recognition of the allelic pair; this association has not heretofore been made.

15           Alternatively, sets of allelic contigs may exist in which the polymorphic site is within an ORF, but does not result in an amino acid change among the allelic polypeptides. Thus, in another embodiment, the polymorphic site resides within an ORF and results in an amino acid change, or a frameshift, among the alleles of the allelic set. In the sets of gene products that fall within this group, at least one of the alleles at the  
20   polypeptide level is a known protein. At least one of the remaining allele or alleles in the set, carrying a variant amino acid at the polymorphic site, is a novel polypeptide not heretofore known. The invention resides at least in the recognition of the polymorphic allele as being a variant of the known reference polypeptide.

          Table 1 provides information concerning the allelic sequences. One of the  
25   sequences may be termed a reference polymorphic sequence, and the corresponding second sequence includes the variant SNP at the polymorphic site. Since the reference polypeptide sequence is already known, the Sequence Listing accompanying this application provides only the sequence of the polymorphic allele, while its SEQ ID NO is provided in the Table. A reference to the SEQ ID NO that corresponds to the translated  
30   amino acid sequence is also given. The Table includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and a description of each, are given below.

5           SNPs disclosed in Table 1 were detected by aligning large numbers of sequences  
from genetically diverse sources of publicly available mRNA libraries (Clontech).  
Software designed specifically to look for multiple examples of variant bases differing  
from a consensus sequence was created and deployed. A criteria of a minimum of 2  
occurrences of a sequence differing from the consensus in high quality sequence reads  
10       was used to identify an SNP.

The SNPs described herein may be useful in diagnostic kits, for DNA arrays on  
chips and for other uses that involve hybridization of the SNP.

Specific SNPs may have utility where a disease has already been associated with  
that gene. Examples of possible disease correlations between the claimed SNPs with  
15       members of the genes of each classification are listed below:

### **Amylases**

Amylase is responsible for endohydrolysis of 1,4-alpha-glucosidic linkages in  
oligosaccharides and polysaccharides. Variations in amylase gene may be indicative of  
delayed maturation and of various amylase producing neoplasms and carcinomas.

### **20    Amyloid**

The serum amyloid A (SAA) proteins comprise a family of vertebrate proteins  
that associate predominantly with high density lipoproteins (HDL). The synthesis of  
certain members of the family is greatly increased in inflammation. Prolonged elevation  
of plasma SAA levels, as in chronic inflammation, 15 results in a pathological condition,  
called amyloidosis, which affects the liver, kidney and spleen and which is characterized  
25       by the highly insoluble accumulation of SAA in these tissues. Amyloid selectively  
inhibits insulin-stimulated glucose utilization and glycogen deposition in muscle, while  
not affecting adipocyte glucose metabolism. Deposition of fibrillar amyloid proteins  
intraneuronally, as neurofibrillary tangles, extracellularly, as plaques and in blood  
30       vessels, is characteristic of both Alzheimer's disease and aged Down's syndrome.  
Amyloid deposition is also associated with type II diabetes mellitus.

## 5 **Angiopoeitin**

Members of the angiopoeitin/fibrinogen family have been shown to stimulate the generation of new blood vessels, inhibit the generation of new blood vessels, and perform several roles in blood clotting. This generation of new blood vessels, called angiogenesis, is also an essential step in tumor growth in order for the tumor to get the blood supply it  
10 needs to expand. Variation in these genes may be predictive of any form of heart disease, numerous blood clotting disorders, stroke, hypertension and predisposition to tumor formation and metastasis. In particular, these variants may be predictive of the response to various antihypertensive drugs and chemotherapeutic and anti-tumor agents.

### **Apoptosis-related proteins**

15 Active cell suicide (apoptosis) is induced by events such as growth factor withdrawal and toxins. It is controlled by regulators, which have either an inhibitory effect on programmed cell death (anti-apoptotic) or block the protective effect of inhibitors (pro-apoptotic). Many viruses have found a way of countering defensive apoptosis by encoding their own anti-apoptosis genes preventing their target-cells from  
20 dying too soon. Variants of apoptosis related genes may be useful in formulation of anti-aging drugs.

### **Cadherin, Cyclin, Polymerase, Oncogenes, Histones, Kinases**

Members of the cell division/cell cycle pathways such as cyclins, many transcription factors and kinases, DNA polymerases, histones, helicases and other  
25 oncogenes play a critical role in carcinogenesis where the uncontrolled proliferation of cells leads to tumor formation and eventually metastasis. Variation in these genes may be predictive of predisposition to any form of cancer, from increased risk of tumor formation to increased rate of metastasis. In particular, these variants may be predictive of the response to various chemotherapeutic and anti-tumor agents.

5 **Colony-stimulating factor-related proteins**

Granulocyte/macrophage colony-stimulating factors are cytokines that act in hematopoiesis by controlling the production, differentiation, and function of 2 related white cell populations of the blood, the granulocytes and the monocytes-macrophages.

**Complement-related proteins**

- 10 Complement proteins are immune associated cytotoxic agents, acting in a chain reaction to exterminate target cells to that were opsonized (primed) with antibodies, by forming a membrane attack complex (MAC). The mechanism of killing is by opening pores in the target cell membrane. Variations in 20 complement genes or their inhibitors are associated with many autoimmune disorders. Modified serum levels of complement
- 15 products cause edemas of various tissues, lupus (SLE), vasculitis, glomerulonephritis, renal failure, hemolytic anemia, thrombocytopenia, and arthritis. They interfere with mechanisms of ADCC (antibody dependent cell cytotoxicity), severely impair immune competence and reduce phagocytic ability. Variants of complement genes may also be indicative of type I diabetes mellitus, meningitis neurological disorders such as Nemaline
- 20 myopathy, Neonatal hypotonia, muscular disorders such as congenital myopathy and other diseases.

**Cytochrome**

- The respiratory chain is a key biochemical pathway which is essential to all aerobic cells. There are five different cytochromes involved in the chain. These are heme
- 25 bound proteins which serve as electron carriers. Modifications in these genes may be predictive of ataxia areflexia, dementia and myopathic and neuropathic changes in muscles. Also, association with various types of solid tumors.

**Kinesins**

- Kinesins are tubulin molecular motors that function to transport organelles within
- 30 cells and to move chromosomes along microtubules during cell division. Modifications of

- 5 these genes may be indicative of neurological disorders such as Pick disease of the brain, tuberous sclerosis.

### **Cytokines, Interferon, Interleukin**

- Members of the cytokine families are known for their potent ability to stimulate cell growth and division even at low concentrations. Cytokines such as erythropoietin are  
10 cell-specific in their growth stimulation; erythropoietin is useful for the stimulation of the proliferation of erythroblasts. Variants in cytokines may be predictive for a wide variety of diseases, including cancer predisposition.

### **G-protein coupled receptors**

- G-protein coupled receptors (also called R7G) are an extensive group of  
15 hormones, neurotransmitters, odorants and light receptors which transduce extracellular signals by interaction with guanine nucleotide-binding (G) proteins. Alterations in genes coding for G-coupled proteins may be involved in and indicative of a vast number of physiological conditions. These include blood pressure regulation, renal dysfunctions, male infertility, dopamine associated cognitive, emotional, and endocrine functions,  
20 hypercalcemia, chondrodysplasia and osteoporosis, pseudohypoparathyroidism, growth retardation and dwarfism.

### **Thioesterases**

- Eukaryotic thiol proteases are a family of proteolytic enzymes which contain an active site cysteine. Catalysis proceeds through a thioester intermediate and is facilitated  
25 by a nearby histidine side chain; an asparagine completes the essential catalytic triad. Variants of thioester associated genes may be predictive of neuronal disorders and mental illnesses such as Ceroid Lipoffiscinosis, Neuronal 1, Infantile, Santavuori disease and more.

### **Breakdown Classifications of SNPS**

- 30 The following list describes the numerical breakdown by molecule type of the SNPs described in Table 1. The key to these molecule types is as follows.



5	TPase_associated:	864
	Guanylyl:	3
	MHC:	1077
	amylase:	44
10	amylaseinhib:	1
	amyloid:	96
	apoptosis:	91
	apoptosisinhib:	29
	apoptosisrecep:	14
15	biotindep:	29
	cadhenn:	415
	calcium_channel:	85
	carboxylase:	4
	cathepsin:	336
20	cathepsininhib:	41
	chloride_channel:	90
	collagen:	1542
	complement:	222
	complementinhib:	21
25	complementrecept:	10
	csf:	31
	csf recept:	37
	cyclin:	65
	cyto45O:	136
30	cytochrome:	659
	deaminase:	44
	dehydrogenase:	1235
	desaturase:	9
	dna_rna_bind:	1309
35	dna_rna_bind_inhib:	16
	dynein:	108
	elastase:	134
	elastaseinhib:	6
	eph:	487
40	esterase:	258
	esteraseinhib:	3
	fgf:	34
	fgf receptor:	12
	gaba:	45
45	glucoamylase:	106
	glucuronidase:	14
	glycoprotein:	3176
	helicase:	333
	histone:	272
50	homeobox:	431
	hydrolase:	187

5	hydroxysteroid:	84
	hypoxanthine:	4
	immunoglob:	1106
	immunoglob_recept:	19
	interferon:	322
10	interleukin:	88
	interleukinrecept:	126
	isomerase:	404
	isomeraseinhibitor:	45
	isomerasereceptor:	4
15	kinase:	1684
	kinase inhibitor:	187
	kinase receptor:	233
	kinesin:	86
	laminin:	196
20	lipase:	63
	metallothionein:	62
	misc_channel:	215
	ngf:	30
	nucl_recpt:	339
25	nuclease:	298
	oncogene:	783
	oxidase:	128
	oxygenase:	14
	peptidase:	150
30	peroxidase:	115
	phosphatase:	668
	phosphataseinhib:	71
	phosphorylase:	84
	polymerase:	489
35	potassium_channel:	43
	prostaglandin:	55
	protease:	954
	proteaseinhib:	271
	reductase:	243
40	ribosomal prot:	1040
	struct:	3128
	sulfotransferase:	42
	synthase:	893
	tgf:	117
45	tgfreceptor:	41
	thioesterase:	3
	thiolase:	38
	tm7:	453
	tnf:	151
50	tnfreceptor:	36

5	traffic:	22
	transcriptfactor:	1139
	transferase:	291
	transport:	900
	tubulin:	334
10	ubiquitin:	229
	water_channel:	18
	unclassified:	10567

The key to the molecule type is as follows:

15

**Abbrev:**

**Title:**

	amylase	amylase protein
	amylaseinhib	amylase inhibitor
20	amyloid	amyloid protein
	apoptosis	apoptosis associated protein
	apoptosisinhib	apoptosis inhibitors
	apoptosisrecep	apoptosis receptors
	ATPase_associated	ATPase associated protein
25	biotindep	biotin dependent enzyme/protein
	cadherin	cadherin protein
	calcium_channel	calcium channel protein
	carboxylase	carboxylase protein
	cathepsin	cathepsin/carboxypeptidases
30	cathepsininhib	cathepsin/carboxypeptidase inhibitor
	chloride_channel	chloride channel protein
	collagen	collagen
	complement	complement protein
	complementrecept	complement receptor protein
35	complementinhib	complement inhibitor
	csf	colony stimulating factor
	csfrecept	colony stimulating factor receptor
	cyclin	cyclin protein
	cyto450	cytochrome p450 protein
40	cytochrome	cytochrome related protein
	deaminase	deaminase
	dehydrogenase	dehydrogenase
	desaturase	desaturase
	dna_rna_bind	DNA/RNA binding protein/factor
45	dna_rna_inhib	DNA/RNA binding protein/factor inhibitor
	dynein	dynein
	elastase	elastase

5	elastaseinhib	elastase inhibitor
	eph	EPH family of tyrosine kinases
	esterase	esterase
	esteraseinhib	esterase inhibitor
	fgf	fibroblast growth factor
10	fgfreceptor	fibroblast growth factor receptor
	gaba	GABA receptor
	glucoamylase	glucoamylase
	glucoronidase	glucoronidase
	glycoprotein	glycoprotein
15	Guanylyl	guanylylate cyclase
	helicase	helicase
	histone	histone
	HOM	homologous
	homeobox	homeobox protein
20	hydrolase	hydrolase
	hydroxysteroid	hydroxysteroid associated protein
	hypoxanthine	hypoxanthine associated protein
	immunoglob	immunoglobulin
	immunoglobrecept	immunoglobulin receptor
25	interferon	interferon
	interleukin	interleukin
	interleukinrecept	interleukin receptor
	isomerase	isomerase
	isomeraseinhibitor	isomerase inhibitor
30	isomerasereceptor	isomerase receptor
	kinase	kinase
	kinaseinhibitor	kinase inhibitor
	kinasereceptor	kinase receptor
	kinesin	kinesin
35	laminin	laminin associated protein
	lipase	lipase
	metallothionein	metallothionein
	MHC	major histocompatibility complex
	misc_channel	miscellaneous channel
40	ngf	nerve growth factor
	nuci_recpt	nuclear receptor
	nuclease	nuclease
	oncogene	oncogene associated protein
	oxidase	oxidase
45	oxygenase	oxygenase
	peptidase	peptidase
	peroxidase	peroxidase
	phosphatase	phosphatase
	phosphataseinhib	phosphatase inhibitor
50	phosphorylase	phosphorylase

5	PIR	PIR DATABASE (release 56, 29-OCT-1998)
	polymerase	polymerase
	potassium_channel	potassium channel protein
	prostaglandin	prostaglandin
10	protease	protease
	proteaseinhib	protease inhibitor
	reductase	reductase
	ribosomalprot	ribosomal associated protein
	RTR	EMBLDATABASE translated
15		entries not to be incorporated into
		SWISS-PROT (20-JUL-1998)
	SIM	similar
	SPTR	EMBL DATABASE translated
20		entries to be incorporated into
		SWISS-PROT (20-JUL-1998)
	struct	structural associated protein
	sulfotransferase	sulfotransferase
	SWP	SWISS-PROT DATABASE (release
		18-OCT-1998)
25	SWPN	SWISS-PROT Update (release 11-NOV-98)
	synthase	synthase
	tgf	transforming growth factor
	tgfreceptor	transforming growth factor receptor
30	thioesterase	thioesterase
	thiolase	thiolase
	tm7	seven transmembrane domain G-
		protein coupled receptor
	tnf	necrosis factor receptor
35	traffic	tumor necrosis factor
	tnfreceptor	tumor trafficking associated protein
	TRN	EMBL DATABASE translated
		entries update (20-JUL-1998)
	transcriptfactor	transcription factor
40	transferase	transferase
	transport	transport protein
	tubulin	tubulin
	ubiquitin	ubiquitin
	unclassified	Protein not categorized into one of
45		the aforementioned protein families
	water channel	water channel protein

Table 1

5           A compilation of polymorphisms is listed in Table 1. Table 1 includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and an explanation for each, are given below.

          The first column of the table lists the names assigned to the fragments in which the polymorphisms occur. The fragments are all human genomic fragments. The  
10   sequence of one allelic form of each of the fragments (arbitrarily referred to as the prototypical or reference form ) has been previously published. These sequences are listed at <http://www-genome.wi.mit.edu/> (all STS's sequence tag sites));  
<http://shgc.stanford.edu> (Stanford STS's); and <http://www.tigr.org/> (TIGR STS's). The web sites also list primers for amplification of the fragments, and the genomic location of  
15   the fragments. Some fragments are expressed sequence tags, and some are random genomic fragments. All information in the web sites concerning the fragments listed in the table is incorporated by reference in its entirety for all purposes.

          The second column lists the position in the fragment in which a polymorphic site has been found. Positions are numbered consecutively with the first base of the fragment  
20   sequence listed as in one of the above databases being assigned the number one. The third column lists the base occupying the polymorphic site in the sequence in the data base. This base is arbitrarily designated the reference or prototypical form, but it is not necessarily the most frequently occurring form. The fourth column in the table lists the alternative base(s) at the polymorphic site. The fifth column of the table lists a 5'  
25   (upstream or forward) primer that hybridizes with the 5' end of the DNA sequence to be amplified. The sixth column of the table lists a 3' (downstream or reverse) primer that hybridizes with the complement of the 3' end of the sequence to be amplified. The seventh column of the table lists a number of bases of sequence on either side of the polymorphic site in each fragment. The indicated sequences can either be DNA or RNA.  
30   In the latter, the T's shown in the table are replaced by U's. The base occupying the polymorphic site is indicated in EUT'AC-IUB ambiguity code.

5           “SEQ ID” provides the cross-references to the two nucleotide SEQ ID NOS: for the cognate pair, which are numbered consecutively, and, as explained below, amino acid SEQ ID NOS: as well, in the Sequence Listing of the application.

Each sequence entry in the Sequence Listing also includes a cross-reference to the CuraGen sequence ID, under the label “Accession number”. The first pair of SEQ ID  
10 NOS: given in the first column of each row of the Table is the SEQ ID NO: identifying the nucleic acid sequence for the polymorphism. If a polymorphism carries an entry for the amino acid portion of the row, a third SEQ ID NO: appears in parentheses in the column “Amino acid before” (see below) for the reference amino acid sequence, and a  
15 fourth SEQ ID NO: appears in parentheses in the column “Amino acid after” (see below) for the polymorphic amino acid sequence. The latter SEQ ID NOS: refer to amino acid sequences giving the cognate reference and polymorphic amino acid sequences that are the translation of the nucleotide polymorphism. If a polymorphism carries no entry for the protein portion of the row, only one pair SEQ ID NOS: is provided, in the first column.

20           “CuraGen sequence ID” provides CuraGen Corporation’s accession number.

“Base pos. of SNP” gives the numerical position of the nucleotide in the nucleic acid at which the cSNP is found, as identified in this invention.

“Polymorphic sequence” provides a 51-base sequence with the polymorphic site at the 26<sup>th</sup> base in the sequence, as well as 25 bases from the reference sequence on the 5’  
25 side and the 3’ side of the polymorphic site. The designation at the polymorphic site is enclosed in square brackets, and provides first, the reference nucleotide; second, a “slash (/)”; and third, the polymorphic nucleotide. In certain cases the polymorphism is an insertion or a deletion. In that case, the position that is “unfilled” (i.e., the reference or the polymorphic position) is indicated by the word “gap”.

30           “Base before” provides the nucleotide present in the reference sequence at the position at which the polymorphism is found.

“Base after” provides the altered nucleotide at the position of the polymorphism.

5           “Amino acid before” provides the amino acid in the reference protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO: in parentheses for the translated reference amino acid sequence if the polymorphism occurs in a coding region.

10           “Amino acid after” provides the amino acid in the polymorphic protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO in parentheses for the translated polymorphic amino acid sequence if the polymorphism occurs in a coding region.

          “Type of change” provides information on the nature of the polymorphism. “SILENT-NONCODING” is used if the polymorphism occurs in a noncoding region of a nucleic acid. “SILENT-CODING” is used if the polymorphism occurs in a coding region  
15           of a nucleic acid of a nucleic acid and results in no change of amino acid in the translated polymorphic protein. “CONSERVATIVE” is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in the same class as the reference amino acid. The classes are: 1) Aliphatic: Gly,  
20           Ala, Val, Leu, Ile; 2) Aromatic: Phe, Tyr, Trp; 3) Sulfur-containing: Cys, Met; 4) Aliphatic OH: Ser, Thr; 5) Basic: Lys, Arg, His; 6) Acidic: Asp, Glu, Asn, Gln; 7) Pro falls in none of the other classes; and 8) End defines a termination codon.

          “NONCONSERVATIVE” is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in a different  
25           class than the reference amino acid.

          “FRAMESHIFT” relates to an insertion or a deletion. If the frameshift occurs in a coding region, the Table provides the translation of the frameshifted codons 3’ to the polymorphic site.

30           “Protein classification of CuraGen gene” provides a generic class into which the protein is classified. Multiple classes of proteins were identified as listed above in the discussion of Table 1.



5           “Name of protein identified following a BLASTX analysis of the CuraGen sequence” provides the database reference for the protein found to resemble the novel reference-polymorphism cognate pair most closely.

          “Similarity (pvalue) following a BLASTX analysis” provides the pvalue, a statistical measure from the BLASTX analysis that the polymorphic sequence is similar  
10 to, and therefore an allele of, the reference, or wild-type, sequence. In the present application, a cutoff of pvalue  $> 1 \times 10^{-50}$  (entered, for example, as 1.0E-50 in the Table) is used to establish that the reference-polymorphic cognate pairs are novel. A pvalue  $< 1 \times 10^{-50}$  defines proteins considered to be already known.

          “Map location” provides any information available at the time of filing related to  
15 localization of a gene on a chromosome.

          The polymorphisms are arranged in Table 1 in the following order:

          SEQ ID NOs: 1-422 are nucleotide sequences for SNPs that are silent.

          SEQ ID NOs: 423-480 are nucleotide sequences for SNPs that lead to conservative amino acid changes.

20           SEQ ID NOs: 481-619 are nucleotide sequences for SNPs that lead to nonconservative amino acid changes.

          SEQ ID NOs: 620-651 are nucleotide sequences for SNPs that involve a gap. With respect to the reference or wild-type sequence at the position of the polymorphism, the allelic cSNP introduces an additional nucleotide (an insertion) or deletes a nucleotide  
25 (a deletion). An SNP that involves a gap generates a frame shift.

          Also presented in the sequence listing filed herewith are predicted amino acid sequences encoded by the polymorphic sequences shown in Table 1.

          SEQ ID NOs: 652-709 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to conservative  
30 amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown.

- 5 The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

SEQ ID NOs: 710-848 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to nonconservative amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

SEQ ID NOs: 849-880 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to frameshift-induced amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

Provided herein are compositions which include, or are capable of detecting, nucleic acid sequences having these polymorphisms, as well as methods of using nucleic acids.

## 20 **Identification of Individuals Carrying SNPs**

Individuals carrying polymorphic alleles of the invention may be detected at either the DNA, the RNA, or the protein level using a variety of techniques that are well known in the art. Strategies for identification and detection are described in *e.g.*, EP 730,663, EP 717,113, and PCT US97/02102. The present methods usually employ pre-characterized polymorphisms. That is, the genotyping location and nature of polymorphic forms present at a site have already been determined. The availability of this information allows sets of probes to be designed for specific identification of the known polymorphic forms.

Many of the methods described below require amplification of DNA from target samples. This can be accomplished by *e.g.*, PCR. (1989), B. for detecting polymorphisms. See generally PCR Technology: Principles and Applications for DNA Amplification (ed. H.A. Erlich, Freeman Press, NY, NY, 1992); PCR Protocols: A

- 5 Guide to Methods and Applications (eds. Innis, et al., Academic Press, San Diego, CA, 1990); Mattila et al., Nucleic Acids Res. 19, 4967 (1991); Eckert et al., PCR Methods and Applications 1, 17 (1991); PCR (eds. McPherson et al., IRL Press, Oxford); and U.S. Patent 4,683,202.

10 The phrase "recombinant protein" or "recombinantly produced protein" refers to a peptide or protein produced using non-native cells that do not have an endogenous copy of DNA able to express the protein. In particular, as used herein, a recombinantly produced protein relates to the gene product of a polymorphic allele, i.e., a "polymorphic protein" containing an altered amino acid at the site of translation of the nucleotide polymorphism. The cells produce the protein because they have been genetically altered  
15 by the introduction of the appropriate nucleic acid sequence. The recombinant protein will not be found in association with proteins and other subcellular components normally associated with the cells producing the protein. The terms "protein" and "polypeptide" are used interchangeably herein.

20 The phrase "substantially purified" or "isolated" when referring to a nucleic acid, peptide or protein, means that the chemical composition is in a milieu containing fewer, or preferably, essentially none, of other cellular components with which it is naturally associated. Thus, the phrase "isolated" or "substantially pure" refers to nucleic acid preparations that lack at least one protein or nucleic acid normally associated with the nucleic acid in a host cell. It is preferably in a homogeneous state although it can be in  
25 either a dry or aqueous solution. Purity and homogeneity are typically determined using analytical chemistry techniques such as gel electrophoresis or high performance liquid chromatography. Generally, a substantially purified or isolated nucleic acid or protein will comprise more than 80% of all macromolecular species present in the preparation. Preferably, the nucleic acid or protein is purified to represent greater than 90% of all  
30 macromolecular species present. More preferably the nucleic acid or protein is purified to greater than 95%, and most preferably the nucleic acid or protein is purified to essential homogeneity, wherein other macromolecular species are not detected by conventional analytical procedures.

5           The genomic DNA used for the diagnosis may be obtained from any nucleated  
cells of the body, such as those present in peripheral blood, urine, saliva, buccal samples,  
surgical specimen, and autopsy specimens. The DNA may be used directly or may be  
amplified enzymatically in vitro through use of PCR (Saiki et al. Science 239:487-491  
(1988)) or other in vitro amplification methods such as the ligase chain reaction (LCR)  
10 (Wu and Wallace Genomics 4:560-569 (1989)), strand displacement amplification (SDA)  
(Walker et al. Proc. Natl. Acad. Sci. U.S.A. 89:392-396 (1992)), self-sustained  
sequence replication (3SR) (Fahy et al. PCR Methods P&J 1:25-33 (1992)), prior to  
mutation analysis.

          The method for preparing nucleic acids in a form that is suitable for mutation  
15 detection is well known in the art. A "nucleic acid" is a deoxyribonucleotide or  
ribonucleotide polymer in either single-or double-stranded form, including known  
analogs of natural nucleotides unless otherwise indicated. The term "nucleic acids", as  
used herein, refers to either DNA or RNA. "Nucleic acid sequence" or "polynucleotide  
sequence" refers to a single-stranded sequence of deoxyribonucleotide or ribonucleotide  
20 bases read from the 5' end to the 3' end. The direction of 5' to 3' addition of nascent  
RNA transcripts is referred to as the transcription direction; sequence regions on the  
DNA strand having the same sequence as the RNA and which are beyond the 5' end of  
the RNA transcript in the 5' direction are referred to as "upstream sequences"; sequence  
regions on the DNA strand having the same sequence as the RNA and which are beyond  
25 the 3' end of the RNA transcript in the 3' direction are referred to as "downstream  
sequences". The term includes both self-replicating plasmids, infectious polymers of  
DNA or RNA and nonfunctional DNA or RNA. The complement of any nucleic acid  
sequence of the invention is understood to be included in the definition of that sequence.  
"Nucleic acid probes" may be DNA or RNA fragments.

30           The detection of polymorphisms in specific DNA sequences, can be accomplished  
by a variety of methods including, but not limited to, restriction-fragment-length-  
polymorphism detection based on allele-specific restriction-endonuclease cleavage (Kan  
and Dozy Lancet ii:910-912 (1978)), hybridization with allele-specific oligonucleotide  
probes (Wallace et al. Nucl. Acids Res. 6:3543-3557 (1978)), including immobilized

5 oligonucleotides (Saiki et al. Proc. Natl. Acad. Sci. USA, 86:6230-6234 (1969)) or  
oligonucleotide arrays (Maskos and Southern Nucl. Acids Res 21:2269-2270 (1993)),  
allele-specific PCR (Newton et al. Nucl Acids Res 17:2503-2516 (1989)), mismatch-  
repair detection (MRD) (Faham and Cox Genome Res 5:474-482 (1995)), binding of  
MutS protein (Wagner et al. Nucl Acids Res 23:3944-3948 (1995)), denaturing-gradient  
10 gel electrophoresis (DGGE) (Fisher and Lerman et al. Proc. Natl. Acad. Sci. U.S.A.  
80:1579-1583 (1983)), single-strand-conformation-polymorphism detection (Orita et al.  
Genomics 5:874-879 (1983)), RNAase cleavage at mismatched base-pairs (Myers et al.  
Science 230:1242 (1985)), chemical (Cotton et al. Proc. Natl. w Sci. U.S.A., 8Z4397-  
4401 (1988)) or enzymatic (Youil et al. Proc. Natl. Acad. Sci. U.S.A. 92:87-91  
15 (1995)) cleavage of heteroduplex DNA, methods based on allele specific primer  
extension (Syvanen et al. Genomics 8:684-692 (1990)), genetic bit analysis (GBA)  
(Nikiforov et al. &&I Acids 22:4167-4175 (1994)), the oligonucleotide-ligation assay  
(OLA) (Landegren et al. Science 241:1077 (1988)), the allele-specific ligation chain  
reaction (LCR) (Barrany Proc. Natl. Acad. Sci. U.S.A. 88:189-193 (1991)), gap-LCR  
20 (Abravaya et al. Nucl Acids Res 23:675-682 (1995)), radioactive and/or fluorescent  
DNA sequencing using standard procedures well known in the art, and peptide nucleic  
acid (PNA) assays (Orum et al., Nucl. Acids Res, 21:5332-5356 (1993); Thiede et al.,  
Nucl. Acids Res. 24:983-984 (1996)).

“Specific hybridization” or “selective hybridization” refers to the binding, or  
25 duplexing, of a nucleic acid molecule only to a second particular nucleotide sequence to  
which the nucleic acid is complementary, under suitably stringent conditions when that  
sequence is present in a complex mixture (e.g., total cellular DNA or RNA). “Stringent  
conditions” are conditions under which a probe will hybridize to its target subsequence,  
but to no other sequences. Stringent conditions are sequence-dependent and are different  
30 in different circumstances. Longer sequences hybridize specifically at higher  
temperatures than shorter ones. Generally, stringent conditions are selected such that the  
temperature is about 5°C lower than the thermal melting point (T<sub>m</sub>) for the specific  
sequence to which hybridization is intended to occur at a defined ionic strength and pH.  
The T<sub>m</sub> is the temperature (under defined ionic strength, pH, and nucleic acid  
35 concentration) at which 50% of the target sequence hybridizes to the complementary

5 probe at equilibrium. Typically, stringent conditions include a salt concentration of at least about 0.01 to about 1.0 M Na ion concentration (or other salts), at pH 7.0 to 8.3. The temperature is at least about 30°C for short probes (e.g., 10 to 50 nucleotides). Stringent conditions can also be achieved with the addition of destabilizing agents such as formamide. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate,  
10 5 mM EDTA, pH 7.4) and a temperature of 25-30°C are suitable for allele-specific probe hybridizations.

“Complementary” or “target” nucleic acid sequences refer to those nucleic acid sequences which selectively hybridize to a nucleic acid probe. Proper annealing conditions depend, for example, upon a probe’s length, base composition, and the number  
15 of mismatches and their position on the probe, and must often be determined empirically. For discussions of nucleic acid probe design and annealing conditions, see, for example, Sambrook et al., or Current Protocols in Molecular Biology, F. Ausubel *et al.*, ed., Greene Publishing and Wiley-Interscience, New York (1987).

A perfectly matched probe has a sequence perfectly complementary to a particular  
20 target sequence. The test probe is typically perfectly complementary to a portion of the target sequence. A “polymorphic” marker or site is the locus at which a sequence difference occurs with respect to a reference sequence. Polymorphic markers include restriction fragment length polymorphisms, variable number of tandem repeats (VNTR's), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats,  
25 tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The reference allelic form may be, for example, the most abundant form in a population, or the first allelic form to be identified, and other allelic forms are designated as alternative, variant or polymorphic alleles. The allelic form occurring most frequently in a selected population is sometimes referred to as the “wild type” form, and herein may also be  
30 referred to as the “reference” form. Diploid organisms may be homozygous or heterozygous for allelic forms. A diallelic polymorphism has two distinguishable forms (i.e., base sequences), and a triallelic polymorphism has three such forms.

As use herein an “oligonucleotide” is a single-stranded nucleic acid ranging in

5 length from 2 to about 60 bases. Oligonucleotides are often synthetic but can also be produced from naturally occurring polynucleotides. A probe is an oligonucleotide capable of binding to a target nucleic acid of complementary sequence through one or more types of chemical bonds, usually through complementary base pairing via hydrogen bond formation. Oligonucleotides probes are often between 5 and 60 bases, and, in  
10 specific embodiments, may be between 10-40, or 15-30 bases long. An oligonucleotide probe may include natural (i.e. A, G, C, or T) or modified bases (7-deazaguanosine, inosine, etc.). In addition, the bases in an oligonucleotide probe may be joined by a linkage other than a phosphodiester bond, such as a phosphoramidite linkage or a phosphorothioate linkage, or they may be peptide nucleic acids in which the constituent  
15 bases are joined by peptide bonds rather than by phosphodiester bonds, so long as it does not interfere with hybridization. Examples of an oligonucleotide are shown in Table 1. Oligonucleotides can be all of a nucleic acid segment as represented in column 4 of Table 1; a nucleic acid sequence which comprises a nucleic acid segment represented in column 4 of Table 1 and additional nucleic acids (present at either or both ends of a nucleic acid  
20 segment of column 4); or a portion (fragment) of a nucleic acid segment represented in column 4 of the table which includes a polymorphic site. Preferred polymorphic sites of the invention include segments of DNA or their complements, which include any one of the polymorphic sites shown in the Table. The segments can be between 5 and 250 bases, and, in specific embodiments are between 5-10, 5-20, 10-20, 10-50, 20-50 or 10-100  
25 bases. The polymorphic site can occur within any position of the segment. The segments can be from any of the allelic forms of the DNA shown in the Table.

As used herein, the term "primer" refers to a single-stranded oligonucleotide which acts as a point of initiation of template-directed DNA synthesis under appropriate conditions (e.g., in the presence of four different nucleoside triphosphates and a  
30 polymerization agent, such as DNA polymerase, RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer, but typically ranges from 15 to 30 nucleotides. Short primer molecules generally require cooler temperatures to form sufficiently stable hybrid complexes with the template. A primer need not be  
35 perfectly complementary to the exact sequence of the template, but should be sufficiently

5 complementary to hybridize with it. The term "primer site" refers to the sequence of the target DNA to which a primer hybridizes. The term "primer pair" refers to a set of primers including a 5' (upstream) primer that hybridizes with the 5' end of the DNA sequence to be amplified and a 3' (downstream) primer that hybridizes with the complement of the 3' end of the sequence to be amplified.

10 DNA fragments can be prepared, for example, by digesting plasmid DNA, or by use of PCR. Oligonucleotides for use as primers or probes are chemically synthesized by methods known in the field of the chemical synthesis of polynucleotides, including by way of non-limiting example the phosphoramidite method described by Beaucage and Carruthers, Tetrahedron Lett 22:1859-1 862 (1981) and the triester method provided by  
15 Matteucci, et al., J. Am. Chem. Soc., 103:3185 (1981) both incorporated herein by reference. These syntheses may employ an automated synthesizer, as described in Needham-VanDevanter, D.R., et al., Nucleic Acids Res. 12:61596168 (1984).  
Purification of oligonucleotides may be carried out by either native acrylamide gel electrophoresis or by anion-exchange HPLC as described in Pearson, J.D. and Regnier,  
20 F.E., J. Chrom., 255:137-149 (1983). A double stranded fragment may then be obtained, if desired, by annealing appropriate complementary single strands together under suitable conditions or by synthesizing the complementary strand using a DNA polymerase with an appropriate primer sequence. Where a specific sequence for a nucleic acid probe is given, it is understood that the complementary strand is also  
25 identified and included. The complementary strand will work equally well in situations where the target is a double-stranded nucleic acid.

The sequence of the synthetic oligonucleotide or of any nucleic acid fragment can be can be obtained using either the dideoxy chain termination method or the Maxam-Gilbert method (see Sambrook et al. Molecular Cloning - a Laboratory Manual (2nd  
30 Ed.), Vols. 1-3, Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, (1989), which is incorporated herein by reference. This manual is hereinafter referred to as "Sambrook et al." ; Zyskind et al., (1988)). Recombinant DNA Laboratory Manual, (Acad. Press, New York). Oligonucleotides useful in diagnostic assays are typically at least 8 consecutive nucleotides in length, and may range upwards of 18 nucleotides in



5 length to greater than 100 or more consecutive nucleotides.

Another aspect of the invention pertains to isolated antisense nucleic acid molecules that are hybridizable to or complementary to the nucleic acid molecule comprising the SNP-containing nucleotide sequences of the invention, or fragments, analogs or derivatives thereof. An "antisense" nucleic acid comprises a nucleotide  
10 sequence that is complementary to a "sense" nucleic acid encoding a protein, *e.g.*, complementary to the coding strand of a double-stranded cDNA molecule or complementary to an mRNA sequence. In specific aspects, antisense nucleic acid molecules are provided that comprise a sequence complementary to at least about 10, about 25, about 50, or about 60 nucleotides or an entire SNP coding strand, or to only a  
15 portion thereof.

In one embodiment, an antisense nucleic acid molecule is antisense to a "coding region" of the coding strand of a polymorphic nucleotide sequence of the invention. The term "coding region" refers to the region of the nucleotide sequence comprising codons which are translated into amino acid. In another embodiment, the antisense nucleic acid  
20 molecule is antisense to a "noncoding region" of the coding strand of a nucleotide sequence of the invention. The term "noncoding region" refers to 5' and 3' sequences which flank the coding region that are not translated into amino acids (*i.e.*, also referred to as 5' and 3' untranslated regions).

Given the coding strand sequences disclosed herein, antisense nucleic acids of the  
25 invention can be designed according to the rules of Watson and Crick or Hoogsteen base pairing. For example, the antisense nucleic acid molecule can generally be complementary to the entire coding region of an mRNA, but more preferably as embodied herein, it is an oligonucleotide that is antisense to only a portion of the coding or noncoding region of the mRNA. An antisense oligonucleotide can range in length  
30 between about 5 and about 60 nucleotides, preferably between about 10 and about 45 nucleotides, more preferably between about 15 and 40 nucleotides, and still more preferably between about 15 and 30 in length. An antisense nucleic acid of the invention can be constructed using chemical synthesis or enzymatic ligation reactions using

5 procedures known in the art. For example, an antisense nucleic acid (*e.g.*, an antisense oligonucleotide) can be chemically synthesized using naturally occurring nucleotides or variously modified nucleotides designed to increase the biological stability of the molecules or to increase the physical stability of the duplex formed between the antisense and sense nucleic acids, *e.g.*, phosphorothioate derivatives and acridine substituted  
10 nucleotides can be used.

Examples of modified nucleotides that can be used to generate the antisense nucleic acid include: 5-fluorouracil, 5-bromouracil, 5-chlorouracil, 5-iodouracil, hypoxanthine, xanthine, 4-acetylcytosine, 5-(carboxyhydroxymethyl) uracil, 5-carboxymethylaminomethyl-2-thiouridine, 5-carboxymethylaminomethyluracil,  
15 dihydrouracil, beta-D-galactosylqueosine, inosine, N6-isopentenyladenine, 1-methylguanine, 1-methylinosine, 2,2-dimethylguanine, 2-methyladenine, 2-methylguanine, 3-methylcytosine, 5-methylcytosine, N6-adenine, 7-methylguanine, 5-methylaminomethyluracil, 5-methoxyaminomethyl-2-thiouracil, beta-D-mannosylqueosine, 5'-methoxycarboxymethyluracil, 5-methoxyuracil,  
20 2-methylthio-N6-isopentenyladenine, uracil-5-oxyacetic acid (v), wybutoxosine, pseudouracil, queosine, 2-thiocytosine, 5-methyl-2-thiouracil, 2-thiouracil, 4-thiouracil, 5-methyluracil, uracil-5-oxyacetic acid methylester, uracil-5-oxyacetic acid (v), 5-methyl-2-thiouracil, 3-(3-amino-3-N-2-carboxypropyl) uracil, (acp3)w, and 2,6-diaminopurine. Alternatively, the antisense nucleic acid can be produced biologically  
25 using an expression vector into which a nucleic acid has been subcloned in an antisense orientation (*i.e.*, RNA transcribed from the inserted nucleic acid will be of an antisense orientation to a target nucleic acid of interest, described further in the following subsection).

The antisense nucleic acid molecules of the invention are typically administered  
30 to a subject or generated *in situ* such that they hybridize with or bind to cellular mRNA and/or genomic DNA encoding a polymorphic protein to thereby inhibit expression of the protein, *e.g.*, by inhibiting transcription and/or translation. The hybridization can be by conventional nucleotide complementary to form a stable duplex, or, for example, in the case of an antisense nucleic acid molecule that binds to DNA duplexes, through specific

5 interactions in the major groove of the double helix. An example of a route of  
administration of antisense nucleic acid molecules of the invention includes direct  
injection at a tissue site. Alternatively, antisense nucleic acid molecules can be modified  
to target selected cells and then administered systemically. For example, for systemic  
administration, antisense molecules can be modified such that they specifically bind to  
10 receptors or antigens expressed on a selected cell surface, *e.g.*, by linking the antisense  
nucleic acid molecules to peptides or antibodies that bind to cell surface receptors or  
antigens. The antisense nucleic acid molecules can also be delivered to cells using the  
vectors described herein. To achieve sufficient intracellular concentrations of antisense  
molecules, vector constructs in which the antisense nucleic acid molecule is placed under  
15 the control of a strong pol II or pol III promoter are preferred.

In yet another embodiment, the antisense nucleic acid molecule of the invention is  
an  $\alpha$ -anomeric nucleic acid molecule. An  $\alpha$ -anomeric nucleic acid molecule forms  
specific double-stranded hybrids with complementary RNA in which, contrary to the usual  
-u n its, the strands run parallel to each other (Gaultier *et al.* (1987) *Nucleic Acids Res* 15:  
20 6625-6641). The antisense nucleic acid molecule can also comprise a  
2'-o-methylribonucleotide (Inoue *et al.* (1987) *Nucleic Acids Res* 15: 6131-6148) or a  
chimeric RNA -DNA analogue (Inoue *et al.* (1987) *FEBS Lett* 215: 327-330).

The following terms are used to describe the sequence relationships between two  
or more nucleic acids or polynucleotides: "reference sequence", "comparison window",  
25 "sequence identity", "percentage of sequence identity", and "substantial identity". A  
"reference sequence" is a defined sequence used as a basis for a sequence comparison; a  
reference sequence may be a subset of a larger sequence, for example, as a segment of a  
full-length cDNA or gene sequence given in a sequence listing, or may comprise a  
complete cDNA or gene sequence. Optimal alignment of sequences for aligning a  
30 comparison window may, for example, be conducted by the local homology algorithm of  
Smith and Waterman Adv. Appl. Math. 2482 (1981), by the homology alignment  
algorithm of Needleman and Wunsch J. Mol. Biol. 48:443 (1970), by the search for  
similarity method of Pearson and Lipman Proc. Natl. Acad. Sci. U.S.A. 852444  
(1988), or by computerized implementations of these algorithms (for example, GAP,

- 5    BESTFIT, FASTA, and TFASTA in the Wisconsin Genetics Software Package Release  
7.0, Genetics Computer Group, 575 Science Dr., Madison, WI).

Techniques for nucleic acid manipulation of the nucleic acid sequences harboring the cSNP's of the invention, such as subcloning nucleic acid sequences encoding polypeptides into expression vectors, labeling probes, DNA hybridization, and the like,  
10    are described generally in Sambrook et al., The phrase "nucleic acid sequence encoding" refers to a nucleic acid which directs the expression of a specific protein, peptide or amino acid sequence. The nucleic acid sequences include both the DNA strand sequence that is transcribed into RNA and the RNA sequence that is translated into protein, peptide or amino acid sequence. The nucleic acid sequences include both the full length nucleic  
15    acid sequences disclosed herein as well as non-full length sequences derived from the full length protein. It being further understood that the sequence includes the degenerate codons of the native sequence or sequences which may be introduced to provide codon preference in a specific host cell. Consequently, the principles of probe selection and array design can readily be extended to analyze more complex polymorphisms (see EP  
20    730,663). For example, to characterize a triallelic SNP polymorphism, three groups of probes can be designed tiled on the three polymorphic forms as described above. As a further example, to analyze a diallelic polymorphism involving a deletion of a nucleotide, one can tile a first group of probes based on the undeleted polymorphic form as the reference sequence and a second group of probes based on the deleted form as the  
25    reference sequence.

For assay of genomic DNA, virtually any biological convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair can be used. Genomic DNA is typically amplified before analysis. Amplification is usually effected by PCR using primers flanking a suitable fragment e.g., of 50-500  
30    nucleotides containing the locus of the polymorphism to be analyzed. Target is usually labeled in the course of amplification. The amplification product can be RNA or DNA, single stranded or double stranded. If double stranded, the amplification product is typically denatured before application to an array. If genomic DNA is analyzed without

5     amplification, it may be desirable to remove RNA from the sample before applying it to the array. Such can be accomplished by digestion with DNase-free RNAase.

#### **DETECTION OF POLYMORPHISMS IN A NUCLEIC ACID SAMPLE**

The SNPs disclosed herein can be used to determine which forms of a characterized polymorphism are present in individuals under analysis.

10           The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki et al., Nature 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the  
15     respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15-  
20     mer at the 7 position; in a 16-mer, at either the 7, 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other member showing a  
25     perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some examples of which are described in published PCT application WO 95/11995.  
30     WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first

5 reference sequence. The second group of probes is designed by the same principles,  
except that the probes exhibit complementarity to the second reference sequence. The  
inclusion of a second group (or further groups) can be particularly useful for analyzing  
short subsequences of the primary reference sequence in which multiple mutations are  
expected to occur within a short distance commensurate with the length of the probes  
10 (e.g., two or more mutations within 9 to 21 bases).

An allele-specific primer hybridizes to a site on target DNA overlapping a  
polymorphism and only primes amplification of an allelic form to which the primer  
exhibits perfect complementarity. See Gibbs, Nucleic Acid Res. 17 2427-2448 (1989).  
This primer is used in conjunction with a second primer which hybridizes at a distal site.  
15 Amplification proceeds from the two-primers, resulting in a detectable product which  
indicates the particular allelic form is present. A control is usually performed with a  
second pair of primers, one of which shows a single base mismatch at the polymorphic  
site and the other of which exhibits perfect complementarity to a distal site. The single-  
base mismatch prevents amplification and no detectable product is formed. The method  
20 works best when the mismatch is included in the 3'-most position of the oligonucleotide  
aligned with the polymorphism because this position is most destabilizing to elongation  
from the primer (see, e.g., WO 93/22456).

Amplification products generated using the polymerase chain reaction can be  
analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can be  
25 identified based on the different sequence-dependent melting properties and  
electrophoretic migration of DNA in solution. Erlich, ed., PCR Technology,  
Principles and Applications for DNA Amplification, (W.H. Freeman and Co New York,  
1992, Chapter 7).

Alleles of target sequences can be differentiated using single-strand conformation  
30 polymorphism analysis, which identifies base differences by alteration in electrophoretic  
migration of single stranded PCR products, as described in Orita et al., Proc. Nat.  
Acad. Sci. 86, 2766-2770 (1989). Amplified PCR products can be generated and  
heated or otherwise denatured, to form single stranded amplification products. Single-

5 stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification products can be related to base-sequence differences between alleles of target sequences.

The genotype of an individual with respect to a pathology suspected of being  
10 caused by a genetic polymorphism may be assessed by association analysis. Phenotypic traits suitable for association analysis include diseases that have known but hitherto unmapped genetic components (e.g., agammaglobulinemia, diabetes insipidus, Lesch-Nyhan syndrome, muscular dystrophy, Wiskott-Aldrich syndrome, Fabry's disease, familial hypercholesterolemia, polycystic kidney disease, hereditary spherocytosis, von  
15 Willebrand's disease, tuberous sclerosis, hereditary hemorrhagic telangiectasia, familial colonic polyposis, Ehlers-Danlos syndrome, osteogenesis imperfecta, and acute intermittent porphyria).

Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases,  
20 inflammation, cancer, system, diseases of the nervous and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid arthritis, multiple sclerosis, diabetes (insulin-dependent and non-independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, brain, breast, colon, esophagus, kidney, oral cavity, ovary, pancreas, prostate,  
25 skin, stomach, leukemia, liver, lung, and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

Such correlations can be exploited in several ways. In the case of a strong  
30 correlation between a polymorphic form and a disease for which treatment is available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple

5 contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be  
10 justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. After determining polymorphic form(s) present in an individual at one or more polymorphic sites, this information can be  
15 used in a number of methods.

Determination of which polymorphic forms occupy a set of polymorphic sites in an individual identifies a set of polymorphic forms that distinguishes the individual. See generally National Research Council, *The Evaluation of Forensic DNA Evidence* (Eds. Pollard et al., National Academy Press, DC, 1996). Since the polymorphic sites are  
20 within a 50,000 bp region in the human genome, the probability of recombination between these polymorphic sites is low. That low probability means the haplotype (the set of all 10 polymorphic sites) set forth in this application should be inherited without change for at least several generations. The more sites that are analyzed the lower the probability that the set of polymorphic forms in one individual is the same as that in an  
25 unrelated individual. Preferably, if multiple sites are analyzed, the sites are unlinked. Thus, polymorphisms of the invention are often used in conjunction with polymorphisms in distal genes. Preferred polymorphisms for use in forensics are diallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

30 The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not



5 match between a suspect and a sample, it can be concluded (barring experimental error)  
that the suspect was not the source of the sample. If the set of markers does match, one  
can conclude that the DNA from the suspect is consistent with that found at the crime  
scene. If frequencies of the polymorphic forms at the loci tested have been determined  
(e.g., by analysis of a suitable population of individuals), one can perform a statistical  
10 analysis to determine the probability that a match of suspect and crime scene sample  
would occur by chance.

$p(ID)$  is the probability that two random individuals have the same polymorphic  
or allelic form at a given polymorphic site. In diallelic loci, four genotypes are possible:  
AA, AB, BA, and BB. If alleles A and B occur in a haploid genome of the organism with  
15 frequencies  $x$  and  $y$ , the probability of each genotype in a diploid organism are (see WO  
95/12607):

$$\text{Homozygote: } p(AA)=x^2$$

$$\text{Homozygote: } p(BB)=y^2=(1-x)^2$$

$$\text{Single Heterozygote: } p(AB)=p(BA)=xy=x(1-x)$$

20  $\text{Both Heterozygotes: } p(AB+BA)=2xy=2x(1-x)$

The probability of identity at one locus (i.e, the probability that two individuals, picked at  
random from a population will have identical polymorphic forms at a given locus) is  
given by the equation:

$$p(ID)=(x^2)^2+(2xy)^2+(y^2)^2.$$

25 These calculations can be extended for any number of polymorphic forms at a  
given locus. For example, the probability of identity  $p(ID)$  for a 3-allele system where the  
alleles have the frequencies in the population of  $x$ ,  $y$  and  $z$ , respectively, is equal to the  
sum of the squares of the genotype frequencies:

$$p(ID)=x^4+(2xy)^2+(2yz)^2+(2xz)^2+z^4+y^4$$

- 5 In a locus of  $n$  alleles, the appropriate binomial expansion is used to calculate  $p(\text{ID})$  and  $p(\text{exc})$ .

The cumulative probability of identity ( $\text{cum } p(\text{ID})$ ) for each of multiple unlinked loci is determined by multiplying the probabilities provided by each locus:

$$\text{cum } p(\text{ID}) = p(\text{ID}1)p(\text{ID}2)p(\text{ID}3) \dots p(\text{ID}n)$$

- 10 The cumulative probability of non-identity for  $n$  loci (i.e. the probability that two random individuals will be different at 1 or more loci) is given by the equation:

$$\text{cum } p(\text{nonID}) = 1 - \text{cum } p(\text{ID}).$$

- If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can  
15 be taken into account together with other evidence in determining the guilt or innocence of the suspect.

- The object of paternity testing is usually to determine whether a male is the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether  
20 the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms in the putative father and the child.

- If the set of polymorphisms in the child attributable to the father does not match the putative father, it can be concluded, barring experimental error, that the putative  
25 father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match.

- The probability of parentage exclusion (representing the probability that a random male will have a polymorphic form at a given polymorphic site that makes him  
30 incompatible as the father) is given by the equation (see WO 95/12607):

5  $p(exc)=xy(1-xy)$

where  $x$  and  $y$  are the population frequencies of alleles A and B of a diallelic polymorphic site. (At a triallelic site  $p(exc)=xy(1-xy)+yz(1-yz)+xz(1-xz)+3xyz(1-xyz)$ ), where  $x$ ,  $y$  and  $z$  are the respective population frequencies of alleles A, B and C). The probability of non-exclusion is:

10  $p(non-exc)=1-p(exc)$

The cumulative probability of non-exclusion (representing the value obtained when  $n$  loci are used) is thus:

$$cum p(non-exc)=p(non-exc1)p(non-exc2)p(non-exc3) \dots p(non-exc n)$$

15 The cumulative probability of exclusion for  $n$  loci (representing the probability that a random male will be excluded) is:

$$cum p(exc)=1-cum p(non-exc).$$

If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the  
20 child's polymorphic marker set attributable to his/her father.

The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending on the  
25 circumstances. For example, a heterozygous sickle cell mutation confers resistance to malaria, but a homozygous sickle cell mutation is usually lethal. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation. A single polymorphism may affect more than one phenotypic trait. Likewise, a single phenotypic trait may be affected by  
30 polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

5           Phenotypic traits include diseases that have known but hitherto unmapped genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid  
10   arthritis, multiple sclerosis, diabetes (insulin-dependent and non-independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, brain, breast, colon, esophagus, kidney, leukemia, liver, lung, oral cavity, ovary, pancreas, prostate, skin, stomach and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed,  
15   endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

          Correlation is performed for a population of individuals who have been tested for the presence or absence of a phenotypic trait of interest and for polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a  
20   polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated with the trait of interest. Correlation can be performed by standard statistical methods such as a  $\chi^2$ -squared test and statistically significant  
25   correlations between polymorphic form(s) and phenotypic characteristics are noted. For example, it might be found that the presence of allele A1 at polymorphism A correlates with heart disease. As a further example, it might be found that the combined presence of allele A1 at polymorphism A and allele B1 at polymorphism B correlates with increased milk production of a farm animal.

30           Such correlations can be exploited in several ways. In the case of a strong correlation between a set of one or more polymorphic forms and a disease for which treatment is available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious

5 disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic  
10 intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. Identification of a polymorphic set in a patient correlated with enhanced  
15 receptiveness to one of several treatment regimes for a disease indicates that this treatment regime should be followed.

For animals and plants, correlations between characteristics and phenotype are useful for breeding for desired characteristics. For example, Beitz et al., U.S. Pat. No. 5,292,639 discuss use of bovine mitochondrial polymorphisms in a breeding program to  
20 improve milk production in cows. To evaluate the effect of mtDNA D-loop sequence polymorphism on milk production, each cow was assigned a value of 1 if variant or 0 if wild type with respect to a prototypical mitochondrial DNA sequence at each of 17 locations considered.

The previous section concerns identifying correlations between phenotypic traits  
25 and polymorphisms that directly or indirectly contribute to those traits. The present section describes identification of a physical linkage between a genetic locus associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Such analysis is useful for mapping a genetic locus associated with a phenotypic  
30 trait to a chromosomal position, and thereby cloning gene(s) responsible for the trait. See Lander et al., *Proc. Natl. Acad. Sci. (USA)* 83, 7353-7357 (1986); Lander et al., *Proc. Natl. Acad. Sci. (USA)* 84, 2363-2367 (1987); Donis-Keller et al., *Cell* 51, 319-337 (1987); Lander et al., *Genetics* 121, 185-199 (1989)). Genes localized by linkage can be cloned by a process known as directional cloning. See Wainwright, *Med. J. Australia*

- 5 159, 170-174 (1993); Collins, *Nature Genetics* 1, 3-6 (1992) (each of which is incorporated by reference in its entirety for all purposes).

Linkage studies are typically performed on members of a family. Available members of the family are characterized for the presence or absence of a phenotypic trait and for a set of polymorphic markers. The distribution of polymorphic markers in an  
10 informative meiosis is then analyzed to determine which polymorphic markers co-segregate with a phenotypic trait. See, e.g., Kerem et al., *Science* 245, 1073-1080 (1989); Monaco et al., *Nature* 316, 842 (1985); Yamoka et al., *Neurology* 40, 222-226 (1990); Rossiter et al., *FASEB Journal* 5, 21-27 (1991).

Linkage is analyzed by calculation of LOD (log of the odds) values. A lod value  
15 is the relative likelihood of obtaining observed segregation data for a marker and a genetic locus when the two are located at a recombination fraction  $r$ , versus the situation in which the two are not linked, and thus segregating independently (Thompson & Thompson, *Genetics in Medicine* (5th ed, W.B. Saunders Company, Philadelphia, 1991); Strachan, "Mapping the human genome" in *The Human Genome* (BIOS Scientific  
20 Publishers Ltd, Oxford), Chapter 4). A series of likelihood ratios are calculated at various recombination fractions ( $r$ ), ranging from  $r=0.0$  (coincident loci) to  $r=0.50$  (unlinked). Thus, the likelihood at a given value of  $r$  is: probability of data if loci linked at  $r$  to probability of data if loci unlinked. The computed likelihood is usually expressed as the  $\log_{10}$  of this ratio (i.e., a lod score). For example, a lod score of 3  
25 indicates 1000:1 odds against an apparent observed linkage being a coincidence. The use of logarithms allows data collected from different families to be combined by simple addition. Computer programs are available for the calculation of lod scores for differing values of  $r$  (e.g., LIPED, MLINK (Lathrop, *Proc. Nat. Acad. Sci. (USA)* 81, 3443-3446 (1984)). For any particular lod score, a recombination fraction may be determined  
30 from mathematical tables. See Smith et al., *Mathematical tables for research workers in human genetics* (Churchill, London, 1961); Smith, *Ann. Hum. Genet.* 32, 127-150 (1968). The value of  $r$  at which the lod score is the highest is considered to be the best estimate of the recombination fraction.

5           Positive lod score values suggest that the two loci are linked, whereas negative  
values suggest that linkage is less likely (at that value of       ) than the possibility that the  
two loci are unlinked. By convention, a combined lod score of + 3 or greater (equivalent  
to greater than 1000:1 odds in favor of linkage) is considered definitive evidence that two  
loci are linked. Similarly, by convention, a negative lod score of -2 or less is taken as  
10   definitive evidence against linkage of the two loci being compared. Negative linkage data  
are useful in excluding a chromosome or a segment thereof from consideration. The  
search focuses on the remaining non-excluded chromosomal locations.

          The invention further provides transgenic nonhuman animals capable of  
expressing an exogenous variant gene and/or having one or both alleles of an endogenous  
15   variant gene inactivated. Expression of an exogenous variant gene is usually achieved  
by operably linking the gene to a promoter and optionally an enhancer, and  
microinjecting the construct into a zygote. See Hogan et al., "Manipulating the Mouse  
Embryo, A Laboratory Manual," Cold Spring Harbor Laboratory. (1989). Inactivation  
of endogenous variant genes can be achieved by forming a transgene in which a cloned  
20   variant gene is inactivated by insertion of a positive selection marker. See Capecchi,  
Science 244, 1288-1292 The transgene is then introduced into an embryonic stem cell,  
where it undergoes homologous recombination with an endogenous variant gene. Mice  
and other rodents are preferred animals. Such animals provide useful drug screening  
systems.

25           The invention further provides methods for assessing the pharmacogenomic  
susceptibility of a subject harboring a single nucleotide polymorphism to a particular  
pharmaceutical compound, or to a class of such compounds. Genetic polymorphism in  
drug-metabolizing enzymes, drug transporters, receptors for pharmaceutical agents, and  
other drug targets have been correlated with individual differences based on distinction in  
30   the efficacy and toxicity of the pharmaceutical agent administered to a subject.  
Pharmacogenomic characterization of a subjects susceptibility to a drug enhances the  
ability to tailor a dosing regimen to the particular genetic constitution of the subject,  
thereby enhancing and optimizing the therapeutic effectiveness of the therapy.

5           In cases in which a cSNP leads to a polymorphic protein that is ascribed to be the  
cause of a pathological condition, method of treating such a condition includes  
administering to a subject experiencing the pathology the wild type cognate of the  
polymorphic protein. Once administered in an effective dosing regimen, the wild type  
cognate provides complementation or remediation of the defect due to the polymorphic  
10   protein. The subject's condition is ameliorated by this protein therapy.

          A subject suspected of suffering from a pathology ascribable to a polymorphic  
protein that arises from a cSNP is to be diagnosed using any of a variety of diagnostic  
methods capable of identifying the presence of the cSNP in the nucleic acid, or of the  
cognate polymorphic protein, in a suitable clinical sample taken from the subject. Once  
15   the presence of the cSNP has been ascertained, and the pathology is correctable by  
administering a normal or wild-type gene, the subject is treated with a pharmaceutical  
composition that includes a nucleic acid that harbors the correcting wild-type gene, or a  
fragment containing a correcting sequence of the wild-type gene. Non-limiting examples  
of ways in which such a nucleic acid may be administered include incorporating the wild-  
20   type gene in a viral vector, such as an adenovirus or adeno associated virus, and  
administration of a naked DNA in a pharmaceutical composition that promotes  
intracellular uptake of the administered nucleic acid. Once the nucleic acid that includes  
the gene coding for the wild-type allele of the polymorphism is incorporated within a cell  
of the subject, it will initiate *de novo* biosynthesis of the wild-type gene product. If the  
25   nucleic acid is further incorporated into the genome of the subject, the treatment will  
have long-term effects, providing *de novo* synthesis of the wild-type protein for a  
prolonged duration. The synthesis of the wild-type protein in the cells of the subject will  
contribute to a therapeutic enhancement of the clinical condition of the subject.

          A subject suffering from a pathology ascribed to a SNP may be treated so as to  
30   correct the genetic defect. (See Kren et al., Proc. Natl. Acad. Sci. USA 96:10349-10354  
(1999)). Such a subject is identified by any method that can detect the polymorphism in  
a sample drawn from the subject. Such a genetic defect may be permanently corrected by  
administering to such a subject a nucleic acid fragment incorporating a repair sequence  
that supplies the wild-type nucleotide at the position of the SNP. This site-specific repair



5 sequence encompasses an RNA/DNA oligonucleotide which operates to promote  
endogenous repair of a subject's genomic DNA. Upon administration in an appropriate  
vehicle, such as a complex with polyethylenimine or encapsulated in anionic liposomes, a  
genetic defect leading to an inborn pathology may be overcome, as the chimeric  
oligonucleotides induces incorporation of the wild-type sequence into the subject's  
10 genome. Upon incorporation, the wild-type gene product is expressed, and the  
replacement is propagated, thereby engendering a permanent repair.

The invention further provides kits comprising at least one allele-specific  
oligonucleotide as described above. Often, the kits contain one or more pairs of allele-  
specific oligonucleotides hybridizing to different forms of a polymorphism. In some  
15 kits, the allele-specific oligonucleotides are provided immobilized to a substrate. For  
example, the same substrate can comprise allele-specific oligonucleotide probes for  
detecting at least 10, 100, 1000 or all of the polymorphisms shown in the Table. Optional  
additional components of the kit include, for example, restriction enzymes, reverse-  
transcriptase or polymerase, the substrate nucleoside triphosphates, means used to label  
20 (for example, an avidin-enzyme conjugate and enzyme substrate and chromogen if the  
label is biotin), and the appropriate buffers for reverse transcription, PCR, or  
hybridization reactions. Usually, the kit also contains instructions for carrying out the  
hybridizing methods.

Several aspects of the present invention rely on having available the polymorphic  
25 proteins encoded by the nucleic acids comprising a SNP of the inventions. There are  
various methods of isolating these nucleic acid sequences. For example, DNA is isolated  
from a genomic or cDNA library using labeled oligonucleotide probes having sequences  
complementary to the sequences disclosed herein.

Such probes can be used directly in hybridization assays. Alternatively probes  
30 can be designed for use in amplification techniques such as PCR.

To prepare a cDNA library, mRNA is isolated from tissue such as heart or  
pancreas, preferably a tissue wherein expression of the gene or gene family is likely to  
occur. cDNA is prepared from the mRNA and ligated into a recombinant vector. The

- 5 vector is transfected into a recombinant host for propagation, screening and cloning. Methods for making and screening cDNA libraries are well known, See Gubler, U. and Hoffman, B.J. Gene 25:263-269 (1983) and Sambrook et al.

For a genomic library, for example, the DNA is extracted from tissue and either mechanically sheared or enzymatically digested to yield fragments of about 12-20 kb.

- 10 The fragments are then separated by gradient centrifugation from undesired sizes and are constructed in bacteriophage lambda vectors. These vectors and phage are packaged *in vitro*, as described in Sambrook, et al. Recombinant phage are analyzed by plaque hybridization as described in Benton and Davis, Science 196:180-182 (1977). Colony hybridization is carried out as generally described in M. Grunstein et al. Proc. Natl. Acad. Sci. USA. 72:3961-3965 (1975). DNA of interest is identified in either cDNA or genomic libraries by its ability to hybridize with nucleic acid probes, for example on Southern blots, and these DNA regions are isolated by standard methods familiar to those of skill in the art. See Sambrook, et al.

- In PCR techniques, oligonucleotide primers complementary to the two 3' borders  
20 of the DNA region to be amplified are synthesized. The polymerase chain reaction is then carried out using the two primers. See PCR Protocols: a Guide to Methods and Applications (Innis, M, Gelfand, D., Sninsky, J. and White, T., eds.), Academic Press, San Diego (1990). Primers can be selected to amplify the entire regions encoding a full-length sequence of interest or to amplify smaller DNA segments as desired. PCR can be  
25 used in a variety of protocols to isolate cDNA's encoding a sequence of interest. In these protocols, appropriate primers and probes for amplifying DNA encoding a sequence of interest are generated from analysis of the DNA sequences listed herein. Once such regions are PCR-amplified, they can be sequenced and oligonucleotide probes can be prepared from the sequence.

- 30 Once DNA encoding a sequence comprising a cSNP is isolated and cloned, one can express the encoded polymorphic proteins in a variety of recombinantly engineered cells. It is expected that those of skill in the art are knowledgeable in the numerous expression systems available for expression of DNA encoding a sequence of interest. No

- 5 attempt to describe in detail the various methods known for the expression of proteins in prokaryotes or eukaryotes is made here.

In brief summary, the expression of natural or synthetic nucleic acids encoding a sequence of interest will typically be achieved by operably linking the DNA or cDNA to a promoter (which is either constitutive or inducible), followed by incorporation into an expression vector. The vectors can be suitable for replication and integration in either prokaryotes or eukaryotes. Typical expression vectors contain, initiation sequences, transcription and translation terminators, and promoters useful for regulation of the expression of a polynucleotide sequence of interest. To obtain high level expression of a cloned gene, it is desirable to construct expression plasmids which contain, at the minimum, a strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. The expression vectors may also comprise generic expression cassettes containing at least one independent terminator sequence, sequences permitting replication of the plasmid in both eukaryotes and prokaryotes, i.e., shuttle vectors, and selection markers for both prokaryotic and eukaryotic systems. See Sambrook et al.

A variety of prokaryotic expression systems may be used to express the polymorphic proteins of the invention. Examples include *E. coli*, *Bacillus*, *Streptomyces*, and the like.

It is preferred to construct expression plasmids which contain, at the minimum, a strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. Examples of regulatory regions suitable for this purpose in *E. coli* are the promoter and operator region of the *E. coli* tryptophan biosynthetic pathway as described by Yanofsky, C., J. Bacterial. 158:1018-1024 (1984) and the leftward promoter of phage lambda (P<sub>L</sub>) as described by A. I. and Hagen, D., Ann. Rev. Genet. 14:399-445 (1980). The inclusion of selection markers in DNA vectors transformed in *E. coli* is also useful. Examples of such markers include genes specifying resistance to ampicillin, tetracycline, or chloramphenicol. See Sambrook et al. for details concerning selection markers for use in *E. coli*.

5 To enhance proper folding of the expressed recombinant protein, during  
purification from *E. coli*, the expressed protein may first be denatured and then renatured.  
This can be accomplished by solubilizing the bacterially produced proteins in a  
chaotropic agent such as guanidine HCl and reducing all the cysteine residues with a  
reducing agent such as beta-mercaptoethanol. The protein is then renatured, either by  
10 slow dialysis or by gel filtration. See U.S. Patent No. 4,511,503. Detection of the  
expressed antigen is achieved by methods known in the art as radioimmunoassay, or  
Western blotting techniques or immunoprecipitation. Purification from *E. coli* can be  
achieved following procedures such as those described in U.S. Patent No. 4,511,503.

Any of a variety of eukaryotic expression systems such as yeast, insect cell lines,  
15 bird, fish, and mammalian cells, may also be used to express a polymorphic protein of the  
invention. As explained briefly below, a nucleotide sequence harboring a cSNP may be  
expressed in these eukaryotic systems. Synthesis of heterologous proteins in yeast is well  
known. Methods in Yeast Genetics, Sherman, F., et al., Cold Spring Harbor Laboratory,  
(1982) is a well recognized work describing the various methods available to produce the  
20 protein in yeast. Suitable vectors usually have expression control sequences, such as  
promoters, including 3-phosphoglycerate kinase or other glycolytic enzymes, and an  
origin of replication, termination sequences and the like as desired. For instance, suitable  
vectors are described in the literature (Botstein, et al., *Gene* 8:17-24 (1979); Broach, et  
al., *Gene* 8:121-133 (1979)).

25 Two procedures are used in transforming yeast cells. In one case, yeast cells are  
first converted into protoplasts using zymolyase, lyticase or glucylase, followed by  
addition of DNA and polyethylene glycol (PEG). The PEG-treated protoplasts are then  
regenerated in a 3% agar medium under selective conditions. Details of this procedure  
are given in the papers by J.D. Beggs, *Nature* (London) 275:104-109 (1978); and  
30 Hinnen, A., et al., *Proc. Natl. Acad. Sci. USA*, 75:1929-1933 (1978). The second  
procedure does not involve removal of the cell wall. Instead the cells are treated with  
lithium chloride or acetate and PEG and put on selective plates (Ito, H., et al., *J. Bact.*  
153:163-168 (1983)). cells and applying standard protein isolation techniques to the  
lysates:.

- 5           The purification process can be monitored by using Western blot techniques or radioimmunoassay or other standard techniques. The sequences encoding the proteins of the invention can also be ligated to various immunoassay expression vectors for use in transforming cell cultures of, for instance, mammalian, insect, bird or fish origin. Illustrative of cell cultures useful for the production of the polypeptides are mammalian
- 10   cells. Mammalian cell systems often will be in the form of monolayers of cells although mammalian cell suspensions may also be used. A number of suitable host cell lines capable of expressing intact proteins have been developed in the art, and include the HEK293, BHK21, and CHO cell lines, and various human cells such as COS cell lines, HeLa cells, myeloma cell lines, Jurkat cells, etc. Expression vectors for these cells can
- 15   include expression control sequences, such as an origin of replication, a promoter (e.g., the CMV promoter, a HSV *tk* promoter or *pgk* (phosphoglycerate kinase) promoter), an enhancer (Queen et al. Immunol. Rev. 89:49 (1986)) and necessary processing information sites, such as ribosome binding sites, RNA splice sites, polyadenylation sites (e.g., an SV40 large T Ag poly A addition site), and transcriptional terminator sequences.
- 20           Other animal cells are available, for instance, from the American Type Culture Collection Catalogue of Cell Lines and Hybridomas (7th edition, (1992)). Appropriate vectors for expressing the proteins of the invention in insect cells are usually derived from baculovirus. Insect cell lines include mosquito larvae, silkworm, armyworm, moth and *Drosophila* cell lines such as a Schneider cell line (See Schneider J. Embryol. Exp.
- 25   Morphol., 27:353-365 (1987). As indicated above, the vector, e.g., a plasmid, which is used to transform the host cell, preferably contains DNA sequences to initiate transcription and sequences to control the translation of the protein. These sequences are referred to as expression control sequences. As with yeast, when higher animal host cells are employed, polyadenylation or transcription terminator sequences from known
- 30   mammalian genes need to be incorporated into the vector. An example of a terminator sequence is the polyadenylation sequence from the bovine growth hormone gene. Sequences for accurate splicing of the transcript may also be included. An example of a splicing sequence is the VP1 intron from SV40 (Sprague, J. et al., J. Virol. 45: 773-781 (1983)). Additionally, gene sequences to control replication in the host cell may be
- 35   Saveria-Campo, M., 1985, "Bovine Papilloma virus DNA a Eukaryotic Cloning Vector"

5 in DNA Cloning Vol. II a Practical Approach Ed. D.M. Glover, IRL Press, Arlington,  
Virginia pp. 213-238. The host cells are competent or rendered competent for  
transformation by various means. There are several well-known methods of introducing  
DNA into animal cells. These include: calcium phosphate precipitation, fusion of the  
recipient cells with bacterial protoplasts containing the DNA, treatment of the recipient  
10 cells with liposomes containing the DNA, DEAE dextran, electroporation and micro-  
injection of the DNA directly into the cells.

The transformed cells are cultured by means well known in the art (Biochemical  
Methods in Cell Culture and Virology, Kuchler, R.J., Dowden, Hutchinson and Ross,  
Inc., (1977)). The expressed polypeptides are isolated from cells grown as suspensions or  
15 as monolayers. The latter are recovered by well known mechanical, chemical or  
enzymatic means.

General methods of expressing recombinant proteins are also known and are  
exemplified in R. Kaufman, Methods in Enzymology 185, 537-566 (1990). As defined  
herein "operably linked" refers to linkage of a promoter upstream from a DNA sequence  
20 such that the promoter mediates transcription of the DNA sequence. Specifically,  
"operably linked" means that the isolated polynucleotide of the invention and an  
expression control sequence are situated within a vector or cell in such a way that the  
gene encoding the protein is expressed by a host cell which has been transformed  
(transfected) with the ligated polynucleotide/expression sequence. The term "vector",  
25 refers to viral expression systems, autonomous self-replicating circular DNA (plasmids),  
and includes both expression and nonexpression plasmids.

The term "gene" as used herein is intended to refer to a nucleic acid sequence  
which encodes a polypeptide. This definition includes various sequence polymorphisms,  
mutations, and/or sequence variants wherein such alterations do not affect the function of  
30 the gene product. The term "gene" is intended to include not only coding sequences but  
also regulatory regions such as promoters, enhancers, termination regions and similar  
untranslated nucleotide sequences. The term further includes all introns and other DNA  
sequences spliced from the mRNA transcript, along with variants resulting from

5 alternative splice sites.

A number of types of cells may act as suitable host cells for expression of the protein. Mammalian host cells include, for example, monkey COS cells, Chinese Hamster Ovary (CHO) cells, human kidney 293 cells, human epidermal A43 1 cells, human Co10205 cells, 3T3 cells, CV-1 cells, other transformed primate cell lines,  
10 normal diploid cells, cell strains derived from in vitro culture of primary tissue, primary explants, HeLa cells, mouse L cells, BHK, HL- 60, U937, HaK or Jurkat cells. Alternatively, it may be possible to produce the protein in lower eukaryotes such as yeast or in prokaryotes such as bacteria. Potentially suitable yeast strains include *Saccharomyces cerevisiae*, *Schizosaccharomyces pombe*, *Kluyveromyces* strains,  
15 *Candida* or any yeast strain capable of expressing heterologous proteins. Potentially suitable bacterial strains include *Escherichia coli*, *Bacillus subtilis*, *Salmonella typhimurium*, or any bacterial strain capable of expressing heterologous proteins. If the protein is made in yeast or bacteria, it may be necessary to modify the protein produced therein, for example by phosphorylation or glycosylation of the appropriate sites, in order  
20 to obtain the functional protein.

The protein may also be produced by operably linking the isolated polynucleotide of the invention to suitable control sequences in one or more insect expression vectors, and employing an insect expression system. Materials and methods for baculovirus/insect cell expression systems are commercially available in kit form from,  
25 e.g., Invitrogen, San Diego, California, U.S.A. (the MaxBac© kit), and such methods are well known in the art, as described in Summers and Smith, Texas Agricultural Experiment Station Bulletin No. 1555 (1987), incorporated herein by reference. As used herein, an insect cell capable of expressing a polynucleotide of the present invention is "transformed." The protein of the invention may be prepared by culturing transformed  
30 host cells under culture conditions suitable to express the recombinant protein.

The polymorphic protein of the invention may also be expressed as a product of transgenic animals, e.g., as a component of the milk of transgenic cows, goats, pigs, or sheep which are characterized by somatic or germ cells containing a nucleotide

- 5 sequence encoding the protein. The protein may also be produced by known conventional chemical synthesis. Methods for constructing the proteins of the present invention by synthetic means are known to those skilled in the art.

- The polymorphic proteins produced by recombinant DNA technology may be purified by techniques commonly employed to isolate or purify recombinant proteins.
- 10 Recombinantly produced proteins can be directly expressed or expressed as a fusion protein. The protein is then purified by a combination of cell lysis (e.g., sonication) and affinity chromatography. For fusion products, subsequent digestion of the fusion protein with an appropriate proteolytic enzyme releases the desired polypeptide. The polypeptides of this invention may be purified to substantial purity by standard
- 15 techniques well known in the art, including selective precipitation with such substances as ammonium sulfate, column chromatography, immunopurification methods, and others. See, for instance, R. Scopes, Protein Purification: Principles and Practice, Springer-Verlag: New York (1982), incorporated herein by reference. For example, in an embodiment, antibodies may be raised to the proteins of the invention as described
- 20 herein. Cell membranes are isolated from a cell line expressing the recombinant protein, the protein is extracted from the membranes and immunoprecipitated. The proteins may then be further purified by standard protein chemistry techniques as described above.

- The resulting expressed protein may then be purified from such culture (i.e., from culture medium or cell extracts) using known purification processes, such as gel
- 25 filtration and ion exchange chromatography. The purification of the protein may also include an affinity column containing agents which will bind to the protein; one or more column steps over such affinity resins as concanavalin A-agarose, heparin-Toyopearl@ or Cibacrom blue 3GA Sepharose B; one or more steps involving hydrophobic interaction chromatography using such resins as phenyl ether, butyl ether, or propyl ether; or
- 30 immunoaffinity chromatography. Alternatively, the protein of the invention may also be expressed in a form which will facilitate purification. For example, it may be expressed as a fusion protein, such as those of maltose binding protein (MBP), glutathione-S-transferase (GST) or thioredoxin (TRX). Kits for expression and purification of such fusion proteins are commercially available from New England BioLab (Beverly, MA),



5 Pharmacia (Piscataway, NJ) and InVitrogen, respectively. The protein can also be tagged with an epitope and subsequently purified by using a specific antibody directed to such epitope. One such epitope ("Flag") is commercially available from Kodak (New Haven, CT). Finally, one or more reverse-phase high performance liquid chromatography (RP-HPLC) steps employing hydrophobic RP-HPLC media, e.g., silica gel having pendant  
10 methyl or other aliphatic groups, can be employed to further purify the protein. Some or all of the foregoing purification steps, in various combinations, can also be employed to provide a substantially homogeneous isolated recombinant protein. The protein thus purified is substantially free of other mammalian proteins and is defined in accordance with the present invention as an "isolated protein."

15 The term "antibody" as used herein refers to immunoglobulin molecules and immunologically active portions of immunoglobulin molecules, *i.e.*, molecules that contain an antigen binding site that specifically binds (immunoreacts with) an antigen, such as polymorphic. Such antibodies include, but are not limited to, polyclonal, monoclonal, chimeric, single chain,  $F_{ab}$  and  $F_{(ab)2}$  fragments, and an  $F_{ab}$  expression  
20 library. In a specific embodiment, antibodies to human polymorphic proteins are disclosed.

The phrase "specifically binds to", "immunospecifically binds to" or is "specifically immunoreactive with", an antibody when referring to a protein or peptide, refers to a binding reaction which is determinative of the presence of the protein in the  
25 presence of a heterogeneous population of proteins and other biological materials. Thus, for example, under designated immunoassay conditions, the specified antibodies bind to a particular protein and do not bind in a significant amount to other proteins present in the sample. Specific binding to an antibody under such conditions may require an antibody that is selected for its specificity for a particular protein. Of particular interest in the  
30 present invention is an antibody that binds immunospecifically to a polymorphic protein but not to its cognate wild type allelic protein, or vice versa. A variety of immunoassay formats may be used to select antibodies specifically immunoreactive with a particular protein. For example, solid-phase ELISA immunoassays are routinely used to select monoclonal antibodies specifically immunoreactive with a protein. See Harlow and Lane

- 5 (1988) Antibodies, a Laboratory Manual, Cold Spring Harbor Publications, New York, for a description of immunoassay formats and conditions that can be used to determine specific immunoreactivity.

Polyclonal and/or monoclonal antibodies that immunospecifically bind to polymorphic gene products but not to the corresponding prototypical or "wild-type" gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, Antibodies, A Laboratory Manual, Cold Spring Harbor Press, New York (1988); Goding, Monoclonal antibodies, Principles and Practice (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific immunoreactivity with a variant gene product and lack of immunoreactivity to the corresponding prototypical gene product.

An isolated polymorphic protein, or a portion or fragment thereof, can be used as an immunogen to generate the antibody that bind the polymorphic protein using standard techniques for polyclonal and monoclonal antibody preparation. The full-length polymorphic protein can be used or, alternatively, the invention provides antigenic peptide fragments of polymorphic for use as immunogens. The antigenic peptide of a polymorphic protein of the invention comprises at least 8 amino acid residues of the amino acid sequence encompassing the polymorphic amino acid and encompasses an epitope of the polymorphic protein such that an antibody raised against the peptide forms a specific immune complex with the polymorphic protein. Preferably, the antigenic peptide comprises at least 10 amino acid residues, more preferably at least 15 amino acid residues, even more preferably at least 20 amino acid residues, and most preferably at least 30 amino acid residues. Preferred epitopes encompassed by the antigenic peptide are regions of polymorphic that are located on the surface of the protein, *e.g.*, hydrophilic regions.

For the production of polyclonal antibodies, various suitable host animals (*e.g.*, rabbit, goat, mouse or other mammal) may be immunized by injection with the polymorphic protein. An appropriate immunogenic preparation can contain, for example,

5 recombinantly expressed polymorphic protein or a chemically synthesized polymorphic polypeptide. The preparation can further include an adjuvant. Various adjuvants used to increase the immunological response include, but are not limited to, Freund's (complete and incomplete), mineral gels (*e.g.*, aluminum hydroxide), surface active substances (*e.g.*, lysolecithin, pluronic polyols, polyanions, peptides, oil emulsions, dinitrophenol, etc.),  
10 human adjuvants such as *Bacille Calmette-Guerin* and *Corynebacterium parvum*, or similar immunostimulatory agents. If desired, the antibody molecules directed against polymorphic proteins can be isolated from the mammal (*e.g.*, from the blood) and further purified by well known techniques, such as protein A chromatography, to obtain the IgG fraction.

15 The term "monoclonal antibody" or "monoclonal antibody composition", as used herein, refers to a population of antibody molecules that originates from the clone of a singly hybridoma cell, and that contains only one type of antigen binding site capable of immunoreacting with a particular epitope of a polymorphic protein. A monoclonal antibody composition thus typically displays a single binding affinity for a particular  
20 polymorphic protein with which it immunoreacts. For preparation of monoclonal antibodies directed towards a particular polymorphic protein, or derivatives, fragments, analogs or homologs thereof, any technique that provides for the production of antibody molecules by continuous cell line culture may be utilized. Such techniques include, but are not limited to, the hybridoma technique (see Kohler & Milstein, 1975 *Nature* 256:  
25 495-497); the trioma technique; the human B-cell hybridoma technique (see Kozbor, *et al.*, 1983 *Immunol Today* 4: 72) and the EBV hybridoma technique to produce human monoclonal antibodies (see Cole, *et al.*, 1985 In: MONOCLONAL ANTIBODIES AND CANCER THERAPY, Alan R. Liss, Inc., pp. 77-96). Human monoclonal antibodies may be utilized in the practice of the present invention and may be produced by using human  
30 hybridomas (see Cote, *et al.*, 1983. *Proc Natl Acad Sci USA* 80: 2026-2030) or by transforming human B-cells with Epstein Barr Virus *in vitro* (see Cole, *et al.*, 1985 In: MONOCLONAL ANTIBODIES AND CANCER THERAPY, Alan R. Liss, Inc., pp. 77-96).

According to the invention, techniques can be adapted for the production of single-chain antibodies specific to a polymorphic protein (see *e.g.*, U.S. Patent No.

- 5 4,946,778). In addition, methodologies can be adapted for the construction of  $F_{ab}$  expression libraries (see *e.g.*, Huse, *et al.*, 1989 *Science* 246: 1275-1281) to allow rapid and effective identification of monoclonal  $F_{ab}$  fragments with the desired specificity for a polymorphic protein or derivatives, fragments, analogs or homologs thereof. Non-human antibodies can be "humanized" by techniques well known in the art. See *e.g.*, U.S. Patent  
10 No. 5,225,539. Antibody fragments that contain the idiotypes to a polymorphic protein may be produced by techniques known in the art including, but not limited to: (i) an  $F_{(ab)2}$  fragment produced by pepsin digestion of an antibody molecule; (ii) an  $F_{ab}$  fragment generated by reducing the disulfide bridges of an  $F_{(ab)2}$  fragment; (iii) an  $F_{ab}$  fragment generated by the treatment of the antibody molecule with papain and a reducing agent  
15 and (iv)  $F_v$  fragments.

Additionally, recombinant anti-polymorphic protein antibodies, such as chimeric and humanized monoclonal antibodies, comprising both human and non-human portions, which can be made using standard recombinant DNA techniques, are within the scope of the invention. Such chimeric and humanized monoclonal antibodies can be produced by  
20 recombinant DNA techniques known in the art, for example using methods described in PCT International Application No. PCT/US86/02269; European Patent Application No. 184,187; European Patent Application No. 171,496; European Patent Application No. 173,494; PCT International Publication No. WO 86/01533; U.S. Pat. No. 4,816,567; European Patent Application No. 125,023; Better *et al.* (1988) *Science* 240:1041-1043;  
25 Liu *et al.* (1987) *PNAS* 84:3439-3443; Liu *et al.* (1987) *J Immunol.* 139:3521-3526; Sun *et al.* (1987) *PNAS* 84:214-218; Nishimura *et al.* (1987) *Cancer Res* 47:999-1005; Wood *et al.* (1985) *Nature* 314:446-449; Shaw *et al.* (1988) *J Natl Cancer Inst* 80:1553-1559; Morrison (1985) *Science* 229:1202-1207; Oi *et al.* (1986) *BioTechniques* 4:214; U.S. Pat. No. 5,225,539; Jones *et al.* (1986) *Nature* 321:552-525; Verhoeyan *et al.* (1988) *Science*  
30 239:1534; and Beidler *et al.* (1988) *J Immunol* 141:4053-4060.

In one embodiment, methodologies for the screening of antibodies that possess the desired specificity include, but are not limited to, enzyme-linked immunosorbent assay (ELISA) and other immunologically-mediated techniques known within the art.

5           Anti-polymorphic protein antibodies may be used in methods known within the art relating to the detection, quantitation and/or cellular or tissue localization of a polymorphic protein (*e.g.*, for use in measuring levels of the polymorphic protein within appropriate physiological samples, for use in diagnostic methods, for use in imaging the protein, and the like). In a given embodiment, antibodies for polymorphic proteins, or  
10 derivatives, fragments, analogs or homologs thereof, that contain the antibody-derived CDR, are utilized as pharmacologically-active compounds in therapeutic applications intended to treat a pathology in a subject that arises from the presence of the cSNP allele in the subject.

          An anti-polymorphic protein antibody (*e.g.*, monoclonal antibody) can be used to  
15 isolate polymorphic proteins by a variety of immunochemical techniques, such as immunoaffinity chromatography or immunoprecipitation. An anti-polymorphic protein antibody can facilitate the purification of natural polymorphic protein from cells and of recombinantly produced polymorphic proteins expressed in host cells. Moreover, an anti-polymorphic protein antibody can be used to detect polymorphic protein (*e.g.*, in a  
20 cellular lysate or cell supernatant) in order to evaluate the abundance and pattern of expression of the polymorphic protein. Anti-polymorphic antibodies can be used diagnostically to monitor protein levels in tissue as part of a clinical testing procedure, *e.g.*, to, for example, determine the efficacy of a given treatment regimen. Detection can be facilitated by coupling (*i.e.*, physically linking) the antibody to a detectable substance.  
25 Examples of detectable substances include various enzymes, prosthetic groups, fluorescent materials, luminescent materials, bioluminescent materials, and radioactive materials. Examples of suitable enzymes include horseradish peroxidase, alkaline phosphatase, -galactosidase, or acetylcholinesterase; examples of suitable prosthetic group complexes include streptavidin/biotin and avidin/biotin; examples of suitable  
30 fluorescent materials include umbelliferone, fluorescein, fluorescein isothiocyanate, rhodamine, dichlorotriazinylamine fluorescein, dansyl chloride or phycoerythrin; an example of a luminescent material includes luminol; examples of bioluminescent materials include luciferase, luciferin, and aequorin, and examples of suitable radioactive material include  $^{125}\text{I}$ ,  $^{131}\text{I}$ ,  $^{35}\text{S}$  or  $^3\text{H}$ .

5

**WHAT IS CLAIMED IS:**

1. An isolated polynucleotide selected from the group consisting of:
  - a) a nucleotide sequence comprising one or more polymorphic sequences (SEQ ID NOS:1 - 651);
  - 10 b) a fragment of said nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence;
  - c) a complementary nucleotide sequence comprising a sequence complementary to one or more of said polymorphic sequences (SEQ ID NOS:1 - 651); and
  - 15 d) a fragment of said complementary nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
2. The polynucleotide of claim 1, wherein said polynucleotide sequence is DNA.
- 20 3. The polynucleotide of claim 1, wherein said polynucleotide sequence is RNA.
4. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 100 nucleotides in length.
- 25 5. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 90 nucleotides in length.

5

6. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 75 nucleotides in length.

7. The polynucleotide of claim 1, wherein said polynucleotide is between about 10  
10 and about 50 bases in length.

8. The polynucleotide of claim 1, wherein said polynucleotide is between about 10  
and about 40 bases in length.

15 9. The polynucleotide of claim 1, wherein said polynucleotide is derived from a  
nucleic acid encoding a polypeptide related to angiopoietin, 4-hydroxybutyrate  
dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility  
antigen, or phosphoglycerate kinase.

20 10. The polynucleotide of claim 1, wherein said polymorphic site includes a  
nucleotide other than the nucleotide listed in Table 1, column 5 for said  
polymorphic sequence.

25 11. The polynucleotide of claim 1, wherein the complement of said polymorphic site  
includes a nucleotide other than the complement of the nucleotide listed in Table  
1, column 5 for the complement of said polymorphic sequence.

- 5      12.      The polynucleotide of claim 1, wherein said polymorphic site includes the  
         nucleotide listed in Table 1, column 6 for said polymorphic sequence.
13.      The polynucleotide of claim 1, wherein the complement of said polymorphic site  
         includes the complement of the nucleotide listed in Table 1, column 6 for said  
10      polymorphic sequence.
14.      An isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide  
         at a polymorphic site encompassed therein, wherein the first polynucleotide is  
         chosen from the group consisting of:
- 15              a) a nucleotide sequence comprising one or more polymorphic sequences  
                 (SEQ ID NOS:1 - 651) provided that the polymorphic sequence  
                 includes a nucleotide other than the nucleotide recited in Table 1,  
                 column 5 for said polymorphic sequence;
- b) a nucleotide sequence that is a fragment of said polymorphic sequence,  
20              provided that the fragment includes a polymorphic site in said  
                 polymorphic sequence;
- c) a complementary nucleotide sequence comprising a sequence  
                 complementary to one or more polymorphic sequences (SEQ ID NOS:1  
                 - 651), provided that the complementary nucleotide sequence includes a  
25              nucleotide other than the complement of the nucleotide recited in Table  
                 1, column 5; and
- d) a nucleotide sequence that is a fragment of said complementary  
                 sequence, provided that the fragment includes a polymorphic site in  
                 said polymorphic sequence.
- 30



- 5      15.      The oligonucleotide of claim 14, wherein the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide selected from the group consisting of:
- 10                      a) a nucleotide sequence comprising one or more polymorphic sequences (SEQ ID NOS:1 - 651), wherein said polymorphic sequence includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence;
- b) a nucleotide sequence that is a fragment of any of said nucleotide sequences;
- 15                      c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences (SEQ ID NOS:1 - 651), wherein said polymorphic sequence includes the complement of the nucleotide listed in Table 1, column 5; and
- d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in
- 20                      said polymorphic sequence.
16.      The oligonucleotide of claim 15, wherein the oligonucleotide is between about 10 and about 51 bases in length.
- 25      17.      The oligonucleotide of claim 15, wherein the oligonucleotide identifies a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

5     18.     The oligonucleotide of claim 15, wherein the oligonucleotide is between about 15  
            and about 30 bases in length.

19.     A method of detecting a polymorphic site in a nucleic acid, the method  
            comprising:

10             a) contacting said nucleic acid with an oligonucleotide that hybridizes to a  
                polymorphic sequence selected from the group consisting of SEQ ID  
                NOS: 1 - 651, or its complement, provided that the polymorphic  
                sequence includes a nucleotide other than the nucleotide recited in  
                Table 1, column 5 for said polymorphic sequence, or the complement  
15             includes a nucleotide other than the complement of the nucleotide  
                recited in Table 1, column 5; and

                b) determining whether said nucleic acid and said oligonucleotide  
                hybridize;

                whereby hybridization of said oligonucleotide to said nucleic acid sequence  
20             indicates the presence of the polymorphic site in said nucleic acid.

20.     The method of claim 19, wherein said oligonucleotide does not hybridize to said  
            polymorphic sequence when said polymorphic sequence includes the nucleotide  
            recited in Table 1, column 5 for said polymorphic sequence, or when the  
25             complement of the polymorphic sequence includes the complement of the  
            nucleotide recited in Table 1, column 5 for said polymorphic sequence.

21.     The method of claim 19, wherein said oligonucleotide identifies a polypeptide  
            related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA  
30             helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

5

22. The method of claim 19, wherein said oligonucleotide is between about 15 and about 30 bases in length.

10

23. A method of detecting the presence of a sequence polymorphism in a subject, the method comprising:

a) providing a nucleic acid from said subject;

15

b) contacting said nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

20

c) determining whether said nucleic acid and said oligonucleotide hybridize;

whereby hybridization of said oligonucleotide to said nucleic acid sequence indicates the presence of the polymorphism in said subject.

25

24. A method of determining the relatedness of a first and second nucleic acid, the method comprising:

a) providing a first nucleic acid and a second nucleic acid;

b) contacting said first nucleic acid and said second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement,

- 5 provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5;
- 10 c) determining whether said first nucleic acid and said second nucleic acid hybridize to said oligonucleotide; and
- d) comparing hybridization of said first and second nucleic acids to said oligonucleotide,
- wherein hybridization of the first and second nucleic acids to said oligonucleotide indicates the first and second nucleic acids are related.

15 25. The method of claim 24, wherein said oligonucleotide does not hybridize to said polymorphic sequence when said polymorphic sequence includes the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the

20 nucleotide recited in Table 1, column 5 for said polymorphic sequence.

26. The method of claim 24, wherein the oligonucleotide is between about 10 and about 51 bases in length.

25 27. The method of claim 24, wherein the oligonucleotide is between about 10 and about 40 bases in length.

28. The method of claim 24, wherein the oligonucleotide is between about 15 and about 30 bases in length.

- 5     29.     An isolated polypeptide comprising a polymorphic site at one or more amino acid  
         residues, wherein the protein is encoded by a polynucleotide selected from the  
         group consisting of: polymorphic sequences SEQ ID NOS:1 - 651, or their  
         complement, provided that the polymorphic sequence includes a nucleotide other  
         than the nucleotide recited in Table 1, column 5 for said polymorphic sequence,  
10           or the complement includes a nucleotide other than the complement of the  
         nucleotide recited in Table 1, column 5.
30.     The polypeptide of claim 29, wherein said polypeptide is translated in the same  
         open reading frame as is a wild type protein whose amino acid sequence is  
15           identical to the amino acid sequence of the polymorphic protein except at the site  
         of the polymorphism.
31.     The polypeptide of claim 29, wherein the polypeptide encoded by said  
         polymorphic sequence, or its complement, includes the nucleotide listed in Table  
20           2, column 6 or Table 3, column 5 for said polymorphic sequence, or the  
         complement includes the complement of the nucleotide listed in Table 1, column  
         6.
32.     An antibody that binds specifically to a polypeptide encoded by a polynucleotide  
25           comprising a nucleotide sequence encoded by a polynucleotide selected from the  
         group consisting of polymorphic sequences SEQ ID NOS:1 - 651, or its  
         complement, provided that the polymorphic sequence includes a nucleotide other  
         than the nucleotide recited in Table 1, column 5 for said polymorphic sequence,  
         or the complement includes a nucleotide other than the complement of the  
30           nucleotide recited in Table 1, column 5.

- 5     33.     The antibody of claim 32, wherein said antibody binds specifically to a  
polypeptide encoded by a polymorphic sequence which includes the nucleotide  
listed in Table 1, column 6 for said polymorphic sequence.
- 10     34.     The antibody of claim 32, wherein said antibody does not bind specifically to a  
polypeptide encoded by a polymorphic sequence which includes the nucleotide  
listed in Table 1, column 5 for said polymorphic sequence.
- 15     35.     A method of detecting the presence of a polypeptide having one or more amino  
acid residue polymorphisms in a subject, the method comprising
- a) providing a protein sample from said subject;
- b) contacting said sample with the antibody of claim 34 under conditions  
         that allow for the formation of antibody-antigen complexes; and
- c) detecting said antibody-antigen complexes,
- whereby the presence of said complexes indicates the presence of said  
20     polypeptide.
36.     A method of treating a subject suffering from, at risk for, or suspected of,  
suffering from a pathology ascribed to the presence of a sequence polymorphism  
in a subject, the method comprising:
- 25     a) providing a subject suffering from a pathology associated with aberrant  
expression of a first nucleic acid comprising a polymorphic sequence  
selected from the group consisting of SEQ ID NOS:1 - 651, or its  
complement; and

- 5                   b) administering to the subject an effective therapeutic dose of a second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide present in a wild type allele of the sequence polymorphism,

thereby treating said subject.

10

37.    The method of claim 36, wherein the second nucleic acid sequence comprises a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence.

- 15    38.   A method of treating a subject suffering from, at risk for, or suspected of suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

- 20                   a) providing a subject suffering from a pathology associated with aberrant expression of a polymorphic sequence selected from the group consisting of polymorphic sequences SEQ ID NOS:1 - 651, or its complement; and

- b) administering to the subject an effective therapeutic dose of a polypeptide,

25                   wherein said polypeptide is encoded by a polynucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence, thereby treating said subject.

30

5 39. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

b) administering to the subject an effective dose of the antibody of claim 34,

thereby treating said subject.

15 40. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

b) administering to the subject an effective dose of an oligonucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence,

thereby treating said subject.



5

41. An oligonucleotide array, comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein, wherein the first polynucleotide is chosen from the group consisting of:

- 10 a) a nucleotide sequence comprising one or more polymorphic sequences  
SEQ ID NOS:1 - 651;
- b) a nucleotide sequence that is a fragment of any of said nucleotide  
sequence, provided that the fragment includes a polymorphic site in  
said polymorphic sequence;
- 15 c) a complementary nucleotide sequence comprising a sequence  
complementary to one or more polymorphic sequences SEQ ID NOS:1  
- 651; and
- d) a nucleotide sequence that is a fragment of said complementary  
sequence, provided that the fragment includes a polymorphic site in  
said polymorphic sequence.

20

42. The array of claim 41, wherein said array comprises 10 oligonucleotides.

43. The array of claim 41, wherein said array comprises at least 100 oligonucleotides.

25 44. The array of claim 41, wherein said array comprises at least 1000  
oligonucleotides.

5

## ABSTRACT

The invention provides nucleic acids containing single-nucleotide polymorphisms identified for transcribed human sequences, as well as methods of using the nucleic acids.

10

15 TRADOCS:1274382.2(rbb\$02!.DOC)

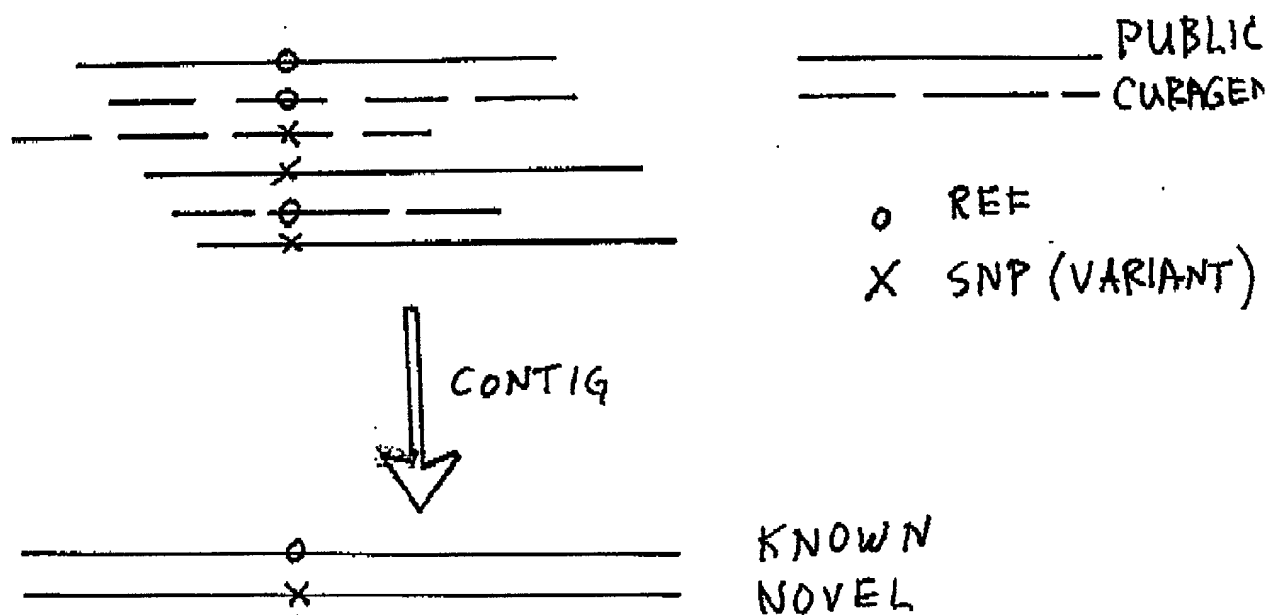


FIG. 1

Attorney Docket No. 15966-534C CIP1  
(CURA-34C CIP1)

## COMBINED DECLARATION AND POWER OF ATTORNEY FOR PATENT APPLICATION

As a below named inventor, I hereby declare that:

My residence, post office address and citizenship are as stated below next to my name.

I believe I am an original, first and joint inventor which is claimed and for which a utility patent is sought on the invention entitled:

# NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE POLYMORPHISMS AND METHODS OF USE THEREOF

the specification of which:

- ☐ was filed on \_\_\_\_\_, as United States non-provisional application  
U.S.S.N. \_\_\_\_\_, bearing Attorney Docket No. \_\_\_\_\_.
- ☒ is attached hereto.

I hereby state that I have reviewed and understand the contents of the above identified specification, including the claims, as amended by any amendment referred to above.

I acknowledge the duty to disclose information which is material to the examination of this application in accordance with Title 37, Code of Federal Regulations, §1.56.

- ☐ I hereby claim foreign priority benefits under Title 35, United States Code, §119(a)-(d) or §365(b) of any foreign application(s) for patent or inventor's certificate, or §365(a) of any PCT International application designating at least one country other than the United States listed below and have also identified below any foreign application for patent or inventor's certificate or PCT International application having a filing date before that of the application on which priority is claimed.

[illegible]

- ☒ I hereby claim the benefit under Title 35, United States Code, § 119(e) or §120 of any United States application(s), or §365(c) of any PCT International application(s) designating the United States of America listed below and, insofar as the subject matter of each of the claims of this application is not disclosed in the prior United States or PCT International application in the manner provided by the first paragraph of Title 35, United States Code, §112, I acknowledge the duty to disclose material information as defined in Title 37, Code of Federal Regulations, §1.56 which became available between the filing date of the prior application and the national or PCT International filing date of this application:

<b>Application No.</b> <i>(U.S.S.N.)</i>	<b>Filing Date</b> <i>(dd/mm/yy)</i>	<b>Status</b> <i>(Patented, Pending, Abandoned)</i>
60/109,024	November 17, 1998	Abandoned
Not Yet Assigned	November 16, 1999	Pending
09/442,129	November 16, 1999	Pending
09/442,849	November 17, 1999	Pending

PCT International Applications designative the United States:

<b>PCT Appln No.</b>	<b>US Serial No.</b>	<b>PCT Filing Date</b>	<b>Status</b>

I hereby appoint the following attorneys and/or agents to prosecute this application and to transact all business in the Patent and Trademark Office connected therewith:

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Kristin E. Konzak	44,848	Shelby J. Walker	45,192
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I hereby declare that all statements made herein of my own knowledge are true and that all statements made on information and belief are believed to be true; and further that these statements were made with the knowledge that willful false statements and the like so made are punishable by fine or imprisonment, or both, under Section 1001 of Title 18 of the United States Code and that such willful false statements may jeopardize the validity of the application or patent issued thereon.

\_\_\_\_\_  
Inventor's Signature

\_\_\_\_\_  
Date

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TRADOCS:1270716.1(R8H\_01!.DOC)

IN THE UNITED STATES PATENT AND TRADEMARK OFFICE

APPLICANTS : Shimkets and Leach  
ASSIGNEE : CuraGen Corporation  
SERIAL NUMBER : Not Yet Assigned EXAMINER : Not Yet Assigned  
FILING DATE : December 27, 1999 ART UNIT : Not Yet Assigned  
FOR : NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE  
POLYMORPHISMS AND METHODS OF USE THEREOF

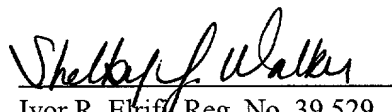
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Washington, D.C. 20231

STATEMENT IN SUPPORT OF COMPUTER READABLE  
FORM SUBMISSION UNDER 37 C.F.R. § 1.821(f)

Sir:

I hereby state that the content of the paper and computer readable forms of the Sequence Listing, submitted in the above-identified application in accordance with 37 C.F.R. § 1.821(c) and 1.821(e), respectively, are the same. No new matter is added.

Respectfully submitted,

  
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**BEFORE THE OFFICE OF ENROLLMENT AND DISCIPLINE  
UNITED STATE PATENT AND TRADEMARK OFFICE**

**LIMITED RECOGNITION UNDER 37 CFR § 10.9(b)**

Michel Morency is hereby given limited recognition under 37 CFR § 10.9(b) as an employee of Mintz, Levin, Cohn, Ferris, Glovsky, and Popeo, P.C. to prepare and prosecute patent applications wherein (1) Michel Morency represents only clients of the Mintz, Levin, Cohn, Ferris, Glovsky, and Popeo, P.C. law firm, and (2) wherein a member of the Mintz, Levin, Cohn, Ferris, Glovsky, and Popeo, P.C. law firm who, in turn, is a registered patent attorney or agent, is the attorney or agent of record in the applications. This limited recognition shall expire on the date appearing below, or when whichever of the following events first occurs prior to the date appearing below: (i) Michel Morency ceases to lawfully reside in the United States, (ii) Michel Morency's employment with Mintz, Levin, Cohn, Ferris, Glovsky, and Popeo, P.C. ceases or is terminated, or (iii) Michel Morency ceases to remain or reside in the United States on H-1B visa.

This document constitutes proof of such recognition. The original of this document is on file in the Office of Enrollment and Discipline of the U.S. Patent and Trademark Office.

**Expires: January 1, 2000**

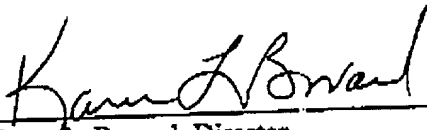
  
\_\_\_\_\_  
Karen L. Bovard, Director  
Office of Enrollment and Discipline



TABLE 1

Seq ID	CuraGen sequence ID	Base pos. of SNP	Polymorphic sequence	Base before	Base after	Amino acid before	Amino acid after	Type of change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	Similarity (pValue) following a BLASTX analysis	Map location
1	cg43333349	1008	CGCTGACAGGGG AGTCTGAGCCACA [A/G]ACCCGCTCA CCCGAGTGCACG CACG	A	G	Gln	Gln	SILENT- CODING	ATPase, as sociated	Human Gene SWISSPROT- ID:P20648 POTASSIUM- TRANSPORTING ATPASE ALPHA CHAIN (EC 3.6.1.36) (PROTON PUMP) (GASTRIC H+/K+ ATPASE ALPHA SUBUNIT) - HOMO SAPIENS (HUMAN), 1035 aa.	0	19
2	cg43931765	2296	ATGGATAGTCCAT CTGGTTGGATGCI A/TJGTGTACTCGT TGGCCTCGTTCAG GT	A	T	Thr	Thr	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3

3	cg44130533	1832	AATACAAAGCTGA GTGGAGAGCAGT T/GJGGTGAAGAAG TATGGCATTCCAA GT	T	G	Val	Val	SILENT- CODING	cadherin	Human Gene SWISSNEW- ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (N-CAM 140) (NCAM-140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa.   pcls:SWISSPROT-ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (N-CAM 140) (NCAM- 140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa.	0	11
4	cg34888922	2330	TATTGTTATTATGT ATTCTGTTTAC/A/ GJTGTTCTGTGTC ACTGCTAAGAGAA	A	G	Thr	Thr	SILENT- CODING	cadherin	Human Gene SWISSNEW- ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2/DG3) - HOMO SAPIENS (HUMAN), 894 aa.   pcls:SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa.	0	18

5	cg34888922	815	CAAGGAGCATTGA CCGTGAGAAATA C/T]GAACAGTTTG CGTTATATGGCTA TG	C	T	Tyr	Tyr	SILENT- CODING	cadherin	Human Gene SWISSNEW- ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2/DG3) - HOMO SAPIENS (HUMAN), 894 aa   pcls:SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa.	0	18
6	cg40310734	1172	TGGCGTCGTATT TGGGCATTTCAGT G/C]GCTGTCACTG ACGTCAACGGGG ATG	G	C	Val	Val	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
7	cg40310734	2243	AGGGGGCCTATG AAGCAGAGCTGG C/C]G]GTGCACCT GCCCCAGGGCGC CCACT	C	G	Ala	Ala	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
8	cg40310734	812	GTTACTGTGAAGC GGCTTCAGCTC C/G]GTGGTCACTC AGGCCGGAGAGC TGG	C	G	Ser	Ser	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)

9	cg43331935	1922	GTGACAAGTACTT CATAGAGGATGG G/TTCGCCTGGTCA TCCACAGCCTGG ACT	G	T	Gly	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P32004 NEURAL CELL ADHESION MOLECULE L1 PRECURSOR (N-CAM L1) - HOMO SAPIENS (HUMAN), 1257 aa.	0	X
10	cg42388009	383	AAGGAGAAACAA TGAAGAACCAGAA C/TJGAAGACGAAG ACTCTGAGGCTGA GA	C	T	Asn	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL- BINDING SIALOPROTEIN) (INTEGRIN-BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	7.00E-172	4
11	cg42388009	389	AAAACAAATGAAGA ACCGAACGAAGA C/TJGAAGACTCTG AGGCTGAGAAATAC CA	C	T	Asp	SILENT- CODING	cadherin	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL- BINDING SIALOPROTEIN) (INTEGRIN-BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	7.00E-172	4
12	cg44126574	1289	AGAACGGCCAGC CCCTGTGGATCCT [C/G]GGGGATGTC TTCCTCAGGTCT ACT	C	G	Leu	SILENT- CODING	cathepsin	Human Gene SPTREMBL- ID:Q64411 PROGASTRICIN PRECURSOR (EC 3.4.23.3) (PEPSIN C) - CAVIA PORCELLUS (GUINEA PIG), 394 aa.	8.00E-155	6 (8p21.3)

13	cg43970983	3066	GGACTCCAGTGT CCAGGGCATCCA G/C/T/TACATCCTA TCCTGGCGGCCA CTCA	C	T	Ser	Ser	SILENT- CODING	collagen	Human Gene SWISSPROT- ID:Q02388 COLLAGEN ALPHA 1(VII) CHAIN PRECURSOR (LONG- CHAIN COLLAGEN) (LC COLLAGEN) - HOMO SAPIENS (HUMAN), 2944 aa.	0	3 (3p21.3)
14	cg44032748	245	TAAGACGGGCAG CTACACCCGCAG C/A/G/GTTACCTG CCAGCTGAGCAA CTGGT	A	G	Ala	Ala	SILENT- CODING	complement	Human Gene SWISSPROT- ID:P07357 COMPLEMENT COMPONENT C8 ALPHA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 584 aa.	0	1 (1p32)
15	cg41553795	222	TCCAGCCCAAGG CCAATTTTGATGC T/G/CAGCAGTTTG CAGGGACCTGGC TCC	T	G	Ala	Ala	SILENT- CODING	complement	Human Gene Homologous to SWISSPROT-ID:P07360 COMPLEMENT C8 GAMMA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 202 aa.	1.40E-104 9 (9q34.3)	
16	cg43942011	1371	AGGTAGGAGGGC TTGGTCTCCAAAC [A/G]CCTATTGTT CATTCTCCACAGT GC	A	G	Gly	Gly	SILENT- CODING	complement recept	Human Gene Similar to TREMBLNEW-ID:E246058 COMPLEMENT RECEPTOR 2 - MUS MUSCULUS (MOUSE), 651 aa (fragment).	1.10E-69	1 (1q32)
17	cg21644442	1219	AACAGCCGGCAG ATGTAACCTGGTAC [A/C]GCCCTTGCCC AGGGTGGGCCCC GTGA	A	C	Thr	Thr	SILENT- CODING	csf	Human Gene SWISSPROT- ID:P09603 MACROPHAGE COLONY STIMULATING FACTOR-1 PRECURSOR (CSF-1) (MCSF) - HOMO SAPIENS (HUMAN), 554 aa.	5.00E-304	1 (1p21)

18	cg41533258	597	TCCAGCGCCGGG CAGGAGGGGTCC TGGAGTTGCCTC CCATCTGCAGAG CTTCC	G	A	Leu	Leu	SILENT- CODING	csf	Human Gene Homologous to SWISSPROT-ID:P09919 GRANULOCYTE COLONY- STIMULATING FACTOR PRECURSOR (G-CSF) (PLURIPROETIN) - HOMO SAPIENS (HUMAN), 207 aa.	1.50E-107	17 (17q11.2)
19	cg43996714	1743	ATGTGCCCACTGC ATTGGTTGTCC A/GGGAGTTGATA CTGGTGGGATCA CAG	A	G	Pro	Pro	SILENT- CODING	dehydrogenase	Human Gene TREMBLNEW- ID:G2979625 PYRUVATE DEHYDROGENASE COMPLEX PROTEIN X SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 501 aa.	1.60E-266	11
20	cg43259523	366	CAGAATATGGAG GCACAGGAGCTT C/A/T/T/T/T/TATCC ACTGTGCTCGTGA TAG	A	T	Ser	Ser	SILENT- CODING	dehydrogenase	Human Gene SWISSPROT- ID:P45954 ACYL-COA DEHYDROGENASE, SHORT/BRANCHED CHAIN SPECIFIC PRECURSOR (EC 1.3.99.-) (SBCAD) (2-METHYL BRANCHED CHAIN ACYL-COA DEHYDROGENASE) (2-MEBCAD) - HOMO SAPIENS (HUMAN), 432 aa.	2.00E-229	10 (10q25)

21	cg43057018	1528	GAATAAGAAATTC AATCTGGATGCAI C/TJTGTTGACCCA TACCCTGCCTTTT GA	C	T	Leu	Leu	SILENT- CODING	dehydrogen ase	Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1) - HOMO SAPIENS (HUMAN), 391 aa [pcls:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.	1.30E-209	4 (4q22)
22	cg1395871	430	GTGCTCCAGAGG GGCCAGCAGGCA C/A/GJGGAAAAAC CGAAACCAACCAA GGACT	A	G	Thr	Thr	SILENT- CODING	dynein	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	2.50E-103	
23	cg1395871	436	CAGAGGGGCCAG CAGGCACAGGAA A/A/GJACCCGAAC CACCAAGGACTTG GCTA	A	G	Lys	Lys	SILENT- CODING	dynein	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	2.50E-103	
24	cg1395871	542	AGCAATGGGAAA GTTTTTTAAAGGA C/TJTGCTTCTTC TGGTGCTTGGGC TTG	C	T	Leu	Leu	SILENT- CODING	dynein	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	2.50E-103	

25	cg1395871	571	CTTCTTCTGGTGC TTGGGCTTGCTT C/TTGATGAATTCA ACCGGATTGAGTT GG	T	Phe	Phe	SILENT- CODING	dynein	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	2.50E-103	
26	cg43950268	1269	AGCGGCCACCA TGGCCCTAGGT C/GATCAACAAGT CCAGCAGCAATCA TGG	A	Asp	Asp	SILENT- CODING	eph	Human Gene TREMBLNEW- ID:G2865466 HEAT SHOCK PROTEIN 75 - HOMO SAPIENS (HUMAN), 649 aa.	0	16
27	cg43918531	461	CCGATGGCTATGA GCAGGCTGCTCG C/TGTGCTATTG AACACCTGGACAA GA	T	Arg	Arg	SILENT- CODING	eph	Human Gene Homologous to SWISSNEW-ID:Q52500 THERMOSOME SUBUNIT (HEAT- SHOCK PROTEIN) - PYROCOCCLUS KODAKARAENSIS, 546 aa.   pcis:SWISSPROT-ID:Q52500 THERMOSOME SUBUNIT (HEAT- SHOCK PROTEIN) - PYROCOCCLUS SP. (STRAIN KOD1), 546 aa.	1.00E-104	5





29	cg43319420	963	TCACCCTCAGGA GGTGGCTGTTCT G/C/TGTCCACGA CAACTACAGAAAC AACC	T	Cys	SILENT- CODING	esterase	Human Gene Similar to SWISSNEW- ID:Q23917 3',5'-CYCLIC- NUCLEOTIDE PHOSPHODIESTERASE REGA (EC 3.1.4.17) (PDEASE REGA) - DICTYOSTELIUM DISCOIDEUM (SLIME MOLD), 793 aa.   pcis:SWISSPROT-ID:Q23917 3',5'-CYCLIC-NUCLEOTIDE PHOSPHODIESTERASE REGA (EC 3.1.4.17) (PDEASE REGA) - DICTYOSTELIUM DISCOIDEUM (SLIME MOLD), 793 aa.	3.30E-60	21
30	cg3001932	1631	TCITCAACATCGT CTATTGGCTTTA[C /T]TATGTGAAC TA AAACATGGCCTCC C	T	Tyr	SILENT- CODING	gaba	Human Gene SWISSPROT- ID:P47870 GAMMA- AMINO BUTYRIC-ACID RECEPTOR BETA-2 SUBUNIT PRECURSOR (GABA(A) RECEPTOR) - HOMO SAPIENS (HUMAN), 474 aa.	1.90E-256	5 (5q34)
31	cg43975899	370	GGATTTTGGACAG ACTCCTAGATGG[ C/T]TATGACAAATC GCCTGAGACCCAG GAT	T	Gly	SILENT- CODING	gaba	Human Gene SWISSPROT- ID:P14867 GAMMA- AMINO BUTYRIC-ACID RECEPTOR ALPHA-1 SUBUNIT PRECURSOR (GABA(A) RECEPTOR) - HOMO SAPIENS (HUMAN), 456 aa.	1.30E-248	5 (5q34)

32	cg43299024	1643	GGGCCCCACTTCC CCCTGGACGTCC A/A/GJTGGAACGA CCTGGACTACATG GACT	A	G	Gln	Gln	SILENT- CODING	glucoamylase	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	7.40E-199	17 (17q25.2)
33	cg43299024	2021	TGAACGAGCCTTC CAACTTCATCAG G/AJGGCTCTGAG GACGGCTGCCCC AACA	G	A	Arg	Arg	SILENT- CODING	glucoamylase	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	7.40E-199	17 (17q25.2)
34	cg43969076	443	AATTCCAAATGAG CTCTCCAACCAC G/AJATTTTCTGC GTTTTTGATCCAG AC	G	A	Tyr	Tyr	SILENT- CODING	glucuronidase	Human Gene SWISSPROT- ID:P08236 BETA-GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA- G1) - HOMO SAPIENS (HUMAN), 651 aa.	0	7 (7q21.11)
35	cg43969014	325	AATTCAGATGAG CTCTCCAACCAC G/AJATTTTCTGC GTTTTTGATCCAG AC	G	A	Tyr	Tyr	SILENT- CODING	glucuronidase	Human Gene Similar to SWISSPROT-ID:P08236 BETA- GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa.	7.40E-80	5
36	cg43065549	880	GGACCATCTCTGT GACCAACCTGCTC G/AJGACGCTGTCA TTGGCCCACTACTC GC	G	A	Ala	Ala	SILENT- CODING	glycoprotein	Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	0	15 (15q15)

37	cg43065549	991	ACCCCTGGAATAG AGAGGATGCTGT T/GJTTCCTGAAGA ATGAGGCTCAGC GCA	T	G	Val	Val	SILENT- CODING	glycoprotein	Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	0	15 (15q15)
38	cg44004239	1141	TGACGTCATCCAT GTCCAATGTCAC C/TIACCAATGCCCC CCCCAAAATGCTC TC	C	T	Val	Val	SILENT- CODING	glycoprotein	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	0	
39	cg44004239	1846	GAAGGGATATAAC TGAAGCAATAAA C/TITTTTCACGGT TGGCAAAATGTGGA CA	C	T	Lys	Lys	SILENT- CODING	glycoprotein	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	0	
40	cg43957605	1677	AGGACTGTTTTTC ATTCAGCTTCAGIA /CJGTGATTCACAT GGGCTCTTCTGTG A	A	C	Thr	Thr	SILENT- CODING	glycoprotein	Human Gene SWISSPROT- ID:Q00013 55 KD ERYTHROCYTE MEMBRANE PROTEIN (P55) - HOMO SAPIENS (HUMAN), 466 aa.	3.10E-249	X (Xq28)

41	cg40915005	1229	ATGTCACAGATT CTACCCAAAGCC C/TGTGTGGTGA TGTGGATGCGGG GTG	C	T	Pro	Pro	SILENT- CODING	glycoprotein	Human Gene SWISSNEW- ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T- CELL SURFACE ANTIGEN T6/LEU- 6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.lpcis:SWISSPROT-ID:P06126 T- CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.	2.00E-183	1 (1q21)
42	cg40356255	1210	TGGCAATAATAGT GCCTTCCTTGCT C/TCTTTTGCTAT GCCTTGCATTATG GT	C	T	Leu	Leu	SILENT- CODING	glycoprotein	Human Gene SWISSNEW- ID:P29016 T-CELL SURFACE GLYCOPROTEIN CD1B PRECURSOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa.lpcis:SWISSPROT-ID:P29016 T- CELL SURFACE GLYCOPROTEIN CD1B PRECURSOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa.	6.70E-183	1 (1q21)





51	cg43983917	1359	ACTTTGCCATTAA CCACAACCCCGA C/TJGCCAAGGACT TGAAGCAGCTCG CGC	C	T	Asp	Asp	SILENT- CODING	homeobox	Human Gene SWISSPROT- ID:P50458 HOMEBOX PROTEIN LH-2 - HOMO SAPIENS (HUMAN), 423 aa.	4.30E-216	
52	cg42730678	979	TGGAGCGAGCGT GGATCCAGTTTCG C/G/JGCGGGGTT GTTGGGTCAAGT TGCT	G	T	Ala	Ala	SILENT- CODING	homeobox	Human Gene SWISSPROT- ID:P28356 HOMEBOX PROTEIN HOX-D9 (HOX-4C) (HOX-5.2) - HOMO SAPIENS (HUMAN), 342 aa.	2.60E-188	2
53	cg42714160	689	GTTACCAGACGCT GGAGCTGGAGAA G/AJGAGTTTCACT ACAATCGCTACCT GA	G	A	Lys	Lys	SILENT- CODING	homeobox	Human Gene Homologous to SWISSPROT-ID:P17509 HOMEBOX PROTEIN HOX-B6 (HOX-2B) (HOX-2.2) (HU-2) - HOMO SAPIENS (HUMAN), 224 aa.	1.10E-123	
54	cg43959084	810	TCAGGTAGCGATT GTAGTGAATTCIT /CJTTCCTCAGCTC CAGGGTCTGGTA GC	T	C	Lys	Lys	SILENT- CODING	homeobox	Human Gene Homologous to SWISSPROT-ID:P09629 HOMEBOX PROTEIN HOX-B7 (HOX-2C) (HHO.C1) - HOMO SAPIENS (HUMAN), 217 aa.	1.30E-113	
55	cg42359655	1124	GGGAAGCATTTG CCAATCAGTCCAG [A/GJGCGGAAAGG GATGCCTTCCTGC AGG	A	G	Arg	Arg	SILENT- CODING	hydrolase	Human Gene SWISSPROT- ID:P09848 LACTASE-PHORIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLKERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa.	0	2 (2q21)



56	cg42359655	2468	ACAGCCAGCGGT TTGGCCTGCACCA [C/T]GTCAACTTCA GCGACAGCAGCA AGT	C	T	His	His	SILENT- CODING	hydrolase	Human Gene SWISSPROT- ID:P09848 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa.	0	2 (2q21)
57	cg42359655	4340	ATCTGGTCACCCCT GCAGAACCTGGG C/T]GTGTCCCACT ACCGTTTTTCCAT CT	C	T	Gly	Gly	SILENT- CODING	hydrolase	Human Gene SWISSPROT- ID:P09848 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa.	0	2 (2q21)
58	cg43998672	1329	TGGTGTGGGCCT TGGTGAACCTCTAG [C/A]ACGCGGCTA ATGCTCCTGGTT TGG	C	A	Val	Val	SILENT- CODING	hydroxyster oid	Human Gene SPTREMBL- ID:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	2.00E-220	16 (16q22)
59	cg43922672	1689	GGAAGCTGACTC CAGAGGCCATGC C/C/T]GACCTCAA CTCCTCCACTGAC TCTG	C	T	Pro	Pro	SILENT- CODING	interleukin	Human Gene TREMBLNEW- ID:G2114410 INTERLEUKIN-16 - HOMO SAPIENS (HUMAN), 631 aa.	0	15

60	cg42908571	630	GTAGTGAGGAAC AAGCCAGAGCTG T[G/C]CAGATGAG TACAAAAGTCCTG ATCC	G	C	Val	Val	SILENT- CODING	interleukin	Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL- 6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.	3.40E-108	7 (7p21)
61	cg43942050	181	AGTTGGAAGTGAA TGGATCGCAGCA C/TTCACCTGACCT GTGCTTTTGAGGA CC	C	T	His	His	SILENT- CODING	interleukinre cept	Human Gene SWISSNEW- ID:P16871 INTERLEUKIN-7 RECEPTOR ALPHA CHAIN PRECURSOR (IL-7R-ALPHA) (CDW127) (CD127 ANTIGEN) - HOMO SAPIENS (HUMAN), 459 aa.   pcds: SWISSPROT-ID: P16871 INTERLEUKIN-7 RECEPTOR ALPHA CHAIN PRECURSOR (IL- 7R-ALPHA) (CDW127) (CD127 ANTIGEN) - HOMO SAPIENS (HUMAN), 459 aa.	3.10E-249	5 (5p13)

62	cg43145505	1249	TAAATATTCGAGA CATTGACAAGATI C/TJTATGTTGAA CAGGTATCTACCA TG	C	T	Ile	Ile	SILENT- CODING	Kinase	Human Gene SWISSNEW- ID:P42336 PHOSPHATIDYLINOSITOL 3- KINASE CATALYTIC SUBUNIT, ALPHA ISOFORM (EC 2.7.1.137) (PI3-KINASE P110 SUBUNIT ALPHA) (PTDINS-3-KINASE P110) (PI3K) - HOMO SAPIENS (HUMAN), 1068 aa.lpcis:SWISSPROT- ID:P42336 PHOSPHATIDYLINOSITOL 3- KINASE CATALYTIC SUBUNIT, ALPHA ISOFORM (EC 2.7.1.137) (PI3-KINASE P110 SUBUNIT ALPHA) (PTDINS-3-KINASE P110) (PI3K) - HOMO SAPIENS (HUMAN), 1068 aa.	0	3
63	cg43918241	1693	AGATCTTTGAGGA AGGGGAATCTGA C/TJGATGAGTTTG ACATGGATGAGAA TC	C	T	Asp	Asp	SILENT- CODING	kinase	Human Gene SPTREMBL- ID:Q63553 SNF1-RELATED KINASE - RATTUS NORVEGICUS (RAT), 746 aa.	0	3
64	cg43090990	1438	TTCTGACGCACAT GTTTGTACATTTC TJTCAGACCAAGGA AAACCTCTTTTTT G	C	T	Phe	Phe	SILENT- CODING	kinase	Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1.-) (NPKC- THETA) - HOMO SAPIENS (HUMAN), 706 aa.	0	10

65	cg43969763	2339	TTAGTATCATTCA CTGTGATCTAAA[A /GJCCTGAAAATAT CCTTCTTTGTAAC C	A	G	Lys	Lys	SILENT- CODING	kinase	Human Gene SWISSPROT- ID:Q13627 SERINE/THREONINE- SPECIFIC PROTEIN KINASE MINIBRAIN HOMOLOG (EC 2.7.1.-) (HP86) (DYRK) - HOMO SAPIENS (HUMAN), 763 aa.	0	21 (21q22.1 )
66	cg42879455	2062	AGGTATATACCAT CATGTACAGTTG[T /CJTGGCATGAGAA AGCAGATGAGCG TC	T	C	Cys	Cys	SILENT- CODING	kinase	Human Gene SWISSPROT- ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) (ATK) (B CELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa.	0	X (Xq21.3)
67	cg42659872	1744	TGGCTCCGGCTA CACCAACATCATG [A/C]GGGTGCTAA GCATATCCTGAGA CGC	A	C	Arg	Arg	SILENT- CODING	kinase	Human Gene SPTREMBL- ID:Q16715 PYRUVATE KINASE (EC 2.7.1.40) - HOMO SAPIENS (HUMAN), 587 aa (fragment).	9.80E-308	1 (1q21)

68	cg42506800	1323	GCTTGCCCAATTTC TCGTCGTGATGCI A/CJAAGTACTTTC AAGGAGATCTGAA TC	A	C	Ala	Ala	SILENT- CODING	Kinase	Human Gene SWISSPROT- ID:Q16654 [PYRUVATE DEHYDROGENASE(LIPOAMIDE)] KINASE ISOZYME 4 PRECURSOR (EC 2.7.1.99) (PYRUVATE DEHYDROGENASE KINASE ISOFORM 4) - HOMO SAPIENS (HUMAN), 411 aa.[pcds:SPTREMBL- ID:Q16654 PYRUVATE DEHYDROGENASE KINASE ISOFORM 4 - HOMO SAPIENS (HUMAN), 411 aa.	1.60E-220	7 (7q21.3)
69	cg43966621	526	CTGTGGAGTACAT GTAGCTGAAGAGI C/TJCGCTCAATCT TCCTCAAGGGAAC AC	C	T	Arg	Arg	SILENT- CODING	Kinase	Human Gene SWISSPROT- ID:Q15119 [PYRUVATE DEHYDROGENASE(LIPOAMIDE)] KINASE ISOZYME 2 PRECURSOR (EC 2.7.1.99) (PYRUVATE DEHYDROGENASE KINASE ISOFORM 2) - HOMO SAPIENS (HUMAN), 407 aa.[pcds:SPTREMBL- ID:Q15119 PYRUVATE DEHYDROGENASE KINASE - HOMO SAPIENS (HUMAN), 407 aa.	3.80E-219	17
70	cg43917871	1448	ACATCATATTGGC GCTGCTGACGGG C/TJGTACTGCCCC CTGGCATGCTAGA TG	C	T	Thr	Thr	SILENT- CODING	Kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)

71	cg43917871	1526	CAGTGTAGAAATA GGGGTGCTCCAT T/GGCCCTCTCTG CAGTAAGCCCGTG ACT	T	G	Ala	Ala	SILENT- CODING	kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)
72	cg44131752	912	AGCTCAATGGTG GCTCTGCGTGCT C/GA/TCCCCGAAG TGACCTGCCTGGT TCCG	G	A	Ser	Ser	SILENT- CODING	kinase	Human Gene SPTREMBL- ID:Q15599 TYROSINE KINASE ACTIVATOR PROTEIN 1 (TKA-1) - HOMO SAPIENS (HUMAN), 450 aa.	7.80E-173	16
73	cg43969473	1765	AATCAACCCACT CATCTATGGCAAT /C/GATGTGGATTC TGTGGATGTTGCA A	T	C	Asn	Asn	SILENT- CODING	kinase	Human Gene SPTREMBL- ID:Q27467 SIMILARITY TO TYROSINE-PROTEIN KINASE - CAENORHABDITIS ELEGANS, 1280 aa.	2.10E-154	11
74	cg44025829	610	AGACCCCGCCGT CCCCTGCCCAAG C/T/C/JGTGGAGTG CTGCCAAGGGGA CTGGT	T	C	Ala	Ala	SILENT- CODING	kinaserecep tor	Human Gene SWISSPROT- ID:Q04771 ACTIVIN RECEPTOR TYPE I PRECURSOR (EC 2.7.1.-) (ACTR-I) (SERINE/THREONINE- PROTEIN KINASE RECEPTOR R1) (SKR1) (ACTIVIN RECEPTOR-LIKE KINASE 2) (ALK-2) (TGF-B SUPERFAMILY RECEPTOR TYPE I) (TSR-I) - HOMO SAPIENS (HUMAN), 509 aa.	7.90E-283	2

75	cg43318277	1107	CTCACGCTTTGCA GTCATCTGGTCC G/AJCCTAGCACTC CCTCCTCTCCTCG GC	G	A	Pro	SILENT- CODING	MHC	Human Gene SPTREMBL- ID:Q02646 MHC BINDING PROTEIN 2 - HOMO SAPIENS (HUMAN), 2500 aa.	1.20E-247	6
76	cg43966144	632	TTAACACGAGGGA GCCTGTGATGCTI G/AJCCCTGCTATG TGTGGGCTTCTA TC	G	A	Leu	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	9.10E-147	6 (6p21.3)
77	cg42686658	644	CCCCTGTGATCAA TATCACCTGGCTI A/GJCGCAACGGC CAAACTGTCACTG AGG	A	G	Leu	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
78	cg42686658	857	CACCACCAGATG CCATGGAGACCC T[G/AJGTCTGTGC CCTGGGCCTGGC CATCG	G	A	Leu	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
79	cg42686658	869	CCATGGAGACCC TGGTCTGTGCCCT [G/AJGGCCTGGCC ATCGGCCTGGTG GGCT	G	A	Leu	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)

80	cg42686658	881	TGGTCTGTGCCCT GGCCTGGCCAT C/TGGCCTGGTG GGCTTCCTCGTG GGCA	C	T	Ile	Ile	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
81	cg42686658	893	TGGGCCTGGCCA TCGGCCTGGTGG G/C/TJTCCTCGT GGGCACCGTCCT CATCA	C	G	Gly	Gly	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
82	cg42686658	905	TCGGCCTGGTGG GCTTCCCTCGTGG G/C/TJACCGTCCT CATCATCATGGGC ACAT	C	T	Gly	Gly	SILENT- CODING	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
83	cg38337333	279	GTTTCCTCATTAG CCCTGTGACCCCI A/TJGCACACGCAG GGACCTACAGAT GTC	A	T	Pro	Pro	SILENT- CODING	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS II INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
84	cg38337333	492	TTGACATCTACCA TCTATCCAGGGA G/AJGGGGAAGCC CATGAACCTTAGGC TCC	G	A	Glu	Glu	SILENT- CODING	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS II INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19



85	cg38337333	699	CTTCTAGTAGTTG GCC TTCACCCAC[ T]A]GAACCAAGCT TCAAAACTGGTAT CG	T	A	Thr	Thr	SILENT- CODING	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
86	cg38337333	774	GGTACTCAGTGG CCATCATCCTCTT[ C]TACCATCCTTC CCTCTTTCTCCT TC	C	T	Phe	Phe	SILENT- CODING	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
87	cg38337333	783	TGGCCATCATCCT CTTCACCATCCTT /C]CCCTTCTTTCT CCTTCATCGCTGG T	T	C	Leu	Leu	SILENT- CODING	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
88	cg43984759	649	AGGAGCTCAAGC GTGAGGCCGAGA C/C/T]CTACGGGA GCGGGAAGGCGA GGAGT	C	T	Thr	Thr	SILENT- CODING	misc_chann el	Human Gene SPTREMBL- ID:Q14193 H-DRK1 K(+) CHANNEL - HOMO SAPIENS (HUMAN), 858 aa.	0	20
89	cg39660131	990	TCATGGGCAACCT AAGGCACAAGTG[ C/T]GTGCGCAACT TCACAGCGCTCAA CG	C	T	Cys	Cys	SILENT- CODING	misc_chann el	Human Gene SPTREMBL- ID:Q14524 SODIUM CHANNEL ALPHA SUBUNIT - HOMO SAPIENS (HUMAN), 2016 aa.	0	3 (3p24)

90	cg44963814	717	CGGAATACCTGG CCATCACCTCTGA [A/G]AGCAAAGAG AACTGCACGGGC GTCC	A	G	Glu	Glu	SILENT- CODING	misc_chann el	Human Gene Homologous to SWISSPROT-ID:Q07699 SODIUM CHANNEL BETA-1 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 218 aa; pcds:TREMBLNEW-ID:G2804300 VOLTAGE-GATED SODIUM CHANNEL BETA-1 SUBUNIT - HOMO SAPIENS (HUMAN), 218 aa.	2.20E-113	19 (19q13.1 )
91	cg21413267	870	AGAGTGGCGAGT GGGTCATCGTGG A/C/TJGCCGTGGG CACCTACAAACACC AGGA	C	T	Asp	Asp	SILENT- CODING	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	7.90E-79	
92	cg21413267	909	ACAAACACCAGGAA GTACGGAGTGCTGJ C/TJGCCGAGATCT ACCCGGACATCA CCT	C	T	Cys	Cys	SILENT- CODING	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	7.90E-79	
93	cg3000465	1160	AGAGGCTCTTTCT GCAGAAACTTCCI A/CJAAATTACTTT GCATGAAAGATCA TG	A	C	Pro	Pro	SILENT- CODING	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	6.10E-70	8 (8p11.2)

94	cg30421838	3766	GTCTAGGATGGA GATCCTACAAACA [C/T]GTCAGTGGG CAGATGCTGTATT TTG	C	T	His	His	SILENT- CODING	nuc_l_recpt	Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.lpcis:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.	0	11 (11q22)
95	cg30421838	4114	ATAACTTGCAATGA TCTTGTCAAACA/A /GJCTTCATCTGTA CTGCTTGAATACA T	A	G	Gln	Gln	SILENT- CODING	nuc_l_recpt	Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.lpcis:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.	0	11 (11q22)
96	cg43947341	713	TTACGTCGCCAAA TTCCAGGGGCACI A/GJTTGCCGACGA ACTTCAGTACGGG AT	A	G	Asn	Asn	SILENT- CODING	nuclease	Human Gene Homologous to SWISSPROT-ID:P07992 DNA EXCISION REPAIR PROTEIN ERCC-1 - HOMO SAPIENS (HUMAN), 297 aa.	1.10E-115	
97	cg43939230	4226	TCCCTGTGACCCA GGCAGGTGCATG A/GJTGACACTGG TCGTGACCTGGC CAG	A	G	Thr	Thr	SILENT- CODING	oncogene	Human Gene SPTREMBL- ID:Q99907 LATENT TRANSFORMING GROWTH FACTOR-BETA-BINDING PROTEIN- 2 - HOMO SAPIENS (HUMAN), 1821 aa.	0	14 (14q24)

98	cg42674136	1447	CGGCACACAGGC CGCTCGCCGGAG C/C/TGTGGCCCA CCCCCAGCCCT GGCCA	C	T	Ala	Ala	SILENT- CODING	oncogene	Human Gene SWISSPROT- ID:P31314 HOMEBOX PROTEIN HOX-11 (TCL-3 PROTO- ONCOGENE) - HOMO SAPIENS (HUMAN), 330 aa.	3.70E-182	10
99	cg41972699	742	AGAACTCGCGGG TCTCCCACTACAT C/TATCAACTCGC TGCCCAACCGCC GTT	C	T	Ile	Ile	SILENT- CODING	oncogene	Human Gene Similar to SWISSPROT-ID:Q64010 PROTO- ONCOGENE C-CRK (P38) (ADAPTER MOLECULE CRK) - MUS MUSCULUS (MOUSE), 304 aa.	2.40E-84	22 (22q11)
100	cg42849556	963	CTGCAACTACCTT GAACCAAGTTGAG C/TTGCGGATCCA CCCTCAGCAGCA GCC	C	T	Leu	Leu	SILENT- CODING	oxidase	Human Gene SWISSPROT- ID:P19878 NEUTROPHIL CYTOSOL FACTOR 2 (NCF-2) (NEUTROPHIL NADPH OXIDASE FACTOR 2) (P67- PHOX) - HOMO SAPIENS (HUMAN), 526 aa.	2.80E-287	1 (1q25)
101	cg43996195	1310	CAGCATGACCTG GCACTGTACTTCG [G/A]GGAAAGTTG GGGATTTCACCGT AGT	G	A	Pro	Pro	SILENT- CODING	phosphoryla se	Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.	2.40E-155	

102	cg43996195	1421	TTGCAACTTGAGG TCGGTGCTTAGT G/AJTGAGACAGAA GCCATTCTGCAGT GT	A	His	SILENT- CODING	phosphoryla se	Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.	2.40E-155	
103	cg43948227	372	TTTACAGTTTCTT ACTGCATCATCIA/ TJATGTCAGAAATC TGTTCCCTTCAGCT	T	Ile	SILENT- CODING	polymerase	Human Gene Similar to SWISSNEW- ID:P53999 ACTIVATED RNA POLYMERASE II TRANSCRIPTIONAL COACTIVATOR P15 (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa.  pcds:SWISSPROT-ID:P53999 ACTIVATED RNA POLYMERASE II TRANSCRIPTIONAL COACTIVATOR P15 (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa.	5.40E-62	5



105	cg43051431	1683	TCACACCTGTCCT GACCCTGGAGGA T/CJGGGTTCTACG AAGTTGACTACAA CA	T	C	Asp	Asp	SILENT- CODING	potassium_ channel	Human Gene SWISSPROT- ID:P48051 G PROTEIN-ACTIVATED INWARD RECTIFIER POTASSIUM CHANNEL 2 (GIRK2) (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 6) (KATP-2) (BIR1) (KIR3.2) - HOMO SAPIENS (HUMAN), 423 aa.lpcds:TREMBLNEW-ID:G1518526 INWARDLY RECTIFYING POTASSIUM CHANNEL KIR3.2 - HOMO SAPIENS (HUMAN), 423 aa.	1.60E-227	16
106	cg43920929	1081	GCAGGATCACCT GCACCCCTCTTGG G/C/GJACCATGAT GCTCATCCAGCTG TCTA	C	G	Val	Val	SILENT- CODING	proteaseinhi b	Human Gene SWISSPROT- ID:P07093 GLIA DERIVED NEXIN PRECURSOR (GDN) (PROTEASE NEXIN I) (PN-1) (PROTEASE INHIBITOR 7) - HOMO SAPIENS (HUMAN), 398 aa.	1.20E-208	2
107	cg43059041	624	AGTCAGACACCA GCTTAGAAATGAC [C/T]ATGGGCAAT GCCTTGTTTCTTG ATG	C	T	Thr	Thr	SILENT- CODING	proteaseinhi b	Human Gene Similar to SWISSPROT-ID:P17475 ALPHA-1- ANTIPROTEINASE PRECURSOR (ALPHA-1-ANTITRYPSIN) (ALPHA- 1- PROTEINASE INHIBITOR) - RATTUS NORVEGICUS (RAT), 411 aa.	4.40E-83	14 (14q32.1 )

108	cg40148056	1385	GGAGGACAGGCA ACTCATCACCGAA [C/T]TAGTCATCAG CAAGATGAACCCAG CT	C	T	Leu	Leu	SILENT- CODING	struct	Human Gene SP TREMBL- ID: Q92777 SYNAPSIN IIB - HOMO SAPIENS (HUMAN), 478 aa.	2.90E-260	3 (3p)
109	cg42894986	1002	ACCCGTTCTTCTG CCCACCCACTGA G/A]GCCCCAGAC CGTGACTTCTTGG TGG	G	A	Glu	Glu	SILENT- CODING	struct	Human Gene SP TREMBL- ID: Q28686 50-KDA DYSTROPHIN- ASSOCIATED GLYCOPROTEIN PRECURSOR - ORYCTOLAGUS CUNICULUS (RABBIT), 387 aa.	1.40E-180	17
110	cg43961212	2160	TCTGGAAGCCGG ACATCCTCTGAGC JA/G]AGTCGACTG ATCCGCTGGCCGA ACCA	A	G	Leu	Leu	SILENT- CODING	struct	Human Gene Homologous to TREMBLNEW-ID: G1703715 PANTOPHYSIN=SYNAPTOPHYSIN HOMOLOG - MUS SP, 261 aa.	2.40E-114	7
111	cg42898003	497	TCATCAGAGATTC GATCTCCTCGTCI C/A]GTCACGTGCT CCCCGGAGGCCCC TGA	C	A	Thr	Thr	SILENT- CODING	struct	Human Gene Similar to SWISSPROT-ID: P02585 TROPONIN C, SKELETAL MUSCLE - HOMO SAPIENS (HUMAN), 159 aa.	1.50E-80	20 (20q12)
112	cg43960684	788	GCTTTGAGGAGG AGGCGCGGTTGC G[C/G]GACGACAC TGAGGCGGCCCAT CCGCG	C	G	Arg	Arg	SILENT- CODING	struct	Human Gene Similar to SWISSPROT-ID: P02535 KERATIN, TYPE I CYTOSKELETAL 10 (CYTOKERATIN 10) (56 KD CYTOKERATIN) (KERATIN, TYPE I CYTOSKELETAL 59 KD) - MUS MUSCULUS (MOUSE), 569 aa.	8.30E-58	8



113	cg43958714	1049	TTCGGAAGGGC AAGCAGTGACCCCT [G/C]ATGATGGAT GCCACCAATATGC CAG	G	C	Leu	Leu	SILENT- CODING	synthase	Human Gene Similar to SPTREMBL- ID:Q42761 SQUALENE SYNTHASE (EC 2.5.1.21) (FARNESYL- DIPHOSPHATE FARNESYLTRANSFERASE) (FARNESYLTRANSFERASE) (PRESQUALENE-DI- DIPHOSPHATE SYNTHASE) - GLYCYRRHIZA GLABRA, 412 aa.	9.20E-83	8
114	cg43124627	901	ACACCCACAGCA GTTTGGTTTAGG A/TTTATCTGTAAA TGGAAGGTTCTG GC	A	T	Gly	Gly	SILENT- CODING	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.   pcds:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.70E-79	16

115	cg43968419	906	TCTTCTCCAACAG TCTGCCACCCGC ATTGTCGTTGGCT GCGCCTCCAAGG CCC	T	Ala	Ala	SILENT- CODING	synthase	Human Gene Similar to SWISSNEW- ID:P53556 8-AMINO-7- OXONONANOATE SYNTHASE (EC 2.3.1.47) (7-KETO-8-AMINO- PELARGONIC ACID SYNTHETASE) (7-KAP SYNTHETASE) (L-ALANINE- -PIMELYL COA LIGASE) - BACILLUS SUBTILIS, 389 aa.  pcis:SWISSPROT-ID:P53556 8- AMINO-7-OXONONANOATE SYNTHASE (EC 2.3.1.47) (7-KETO- 8-AMINO- PELARGONIC ACID SYNTHETASE) (7-KAP SYNTHETASE) (L-ALANINE-- PIMELYL COA LIGASE) - BACILLUS SUBTILIS, 389 aa.	9.90E-70
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116	cg43064068	1484	TTGTGGTCCTGGC CTCGCAGTTCCTI G/AJTCCCATGACC CAGAACAGCTCAC CA	G	A	Leu	Leu	SILENT- CODING	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.lpcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.40E-65	
117	cg43064068	1622	TCACAGGGAATA TCAACGAGCCAAI G/AJCTTCGAGACA AGGAGTGAAGA TGT	G	A	Lys	Lys	SILENT- CODING	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.lpcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.40E-65	

118	cg41084924	1278	TGACTCTCCCCGAC CCCGTCCCACCAI C/TJGGTCTCCACA GCACTCCCGACA GCC	C	T	His	His	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	1.70E-241	11
119	cg41084924	1662	TCCGCAAGGCCTT CCTGAAGATCCTI C/TJCACTGCTGAC TCTGCTGCCTGCC CG	C	T	Leu	Leu	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	1.70E-241	11
120	cg41084924	606	TCCTCGTCGCCAC ACTGGTCATGCCI C/ATGGGTTGTCT ACCTGGAGGTGG TAG	C	A	Pro	Pro	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	1.70E-241	11
121	cg43985000	1471	TTGCTCTTTGCTG GTTCCCTCTTCAIC /TJTTAAGCCGTAT ATTGAAGAAACT G	C	T	His	His	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa.	1.60E-236	4
122	cg43985000	1507	TATTGAAGAAAC TGTGTATAACGAIA /GJATGGACAAGAA CCGATGTGAATTA C	A	G	Glu	Glu	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa.	1.60E-236	4

123	cg44930578	561	ACGTGAACACCG ACATCTACTCCAA G/AJGTGCTGGTGA CCGCCGTGTACC TGG	G	A	Lys	Lys	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P30989 NEUROTENSIN RECEPTOR TYPE 1 (NT-R-1) (HIGH-AFFINITY LEVOCABASTINE- INSENSITIVE NEUROTENSIN RECEPTOR) (NTRH) - HOMO SAPIENS (HUMAN), 418 aa.	5.00E-217	
124	cg3003519	1263	ATTCCTTGATTGC TAGGACCCCTTTA C/TJAAAAGCACCC TGAACATACCTAC TG	C	T	Tyr	Tyr	SILENT- CODING	tm7	Human Gene SWISSNEW- ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.   pcis: SWISSPROT-ID:P32247 BOMBESIN RECEPTOR SUBTYPE- 3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.   pcis: TREMBLNEW-ID:E1240254 BOMBESIN RECEPTOR SUBTYPE- 3 (UTERINE BOMBESIN RECEPTOR, BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.	3.00E-212	X

125	cg3003519	711	C	T	Gly	Gly	SILENT-CODING	tm7	Human Gene SWISSNEW-ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa. aa.pclis:SWISSPROT-ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa. aa.pclis:TREMBLNEW-ID:E1240254 BOMBESIN RECEPTOR SUBTYPE-3 (UTERINE BOMBESIN RECEPTOR, BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.	3.00E-212	X
126	cg43969010	1182	C	T	Tyr	Tyr	SILENT-CODING	tm7	Human Gene SWISSPROT-ID:P30411 B2 BRADYKININ RECEPTOR (BK-2 RECEPTOR) - HOMO SAPIENS (HUMAN), 391 aa.	9.00E-211	12 (14q32.1)
127	cg43263108	1097	C	A	Ser	Ser	SILENT-CODING	tm7	Human Gene SWISSPROT-ID:P43119 PROSTACYCLIN RECEPTOR (PROSTANOID IP RECEPTOR) (PGI RECEPTOR) - HOMO SAPIENS (HUMAN), 386 aa.	8.30E-208	19 (19q13.3)
128	cg43263108	272	C	G	Val	Val	SILENT-CODING	tm7	Human Gene SWISSPROT-ID:P43119 PROSTACYCLIN RECEPTOR (PROSTANOID IP RECEPTOR) (PGI RECEPTOR) - HOMO SAPIENS (HUMAN), 386 aa.	8.30E-208	19 (19q13.3)

129	cg43267238	1220	CCAGACTGGTCCT GGTGGTGGTGGC[ A/G]GTCTTCGTGG TCTGCTGGACTCC CA	A	G	Ala	Ala	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	2.10E-204	8 (8q11.2)
130	cg43267238	392	CAGCACTCACCAT GGAATCCCCGAT[ C/T]CAGATCTTCC GCGGGGAGCCGG GCC	C	T	Ile	Ile	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	2.10E-204	8 (8q11.2)
131	cg43267238	413	CGATCCAGATCTT CCGCGGGGAGCC [G/T]GGCCCTACC TGCGCCCCCGAGC GCCT	G	T	Pro	Pro	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	2.10E-204	8 (8q11.2)
132	cg43264978	155	TGGATCTGCACCT CTTCGACTACTC[A /C]GAGCCAGGGA ACTTCTCGGACAT CA	A	C	Ser	Ser	SILENT- CODING	tm7	Human Gene TREMBLNEW- ID:G2736282 G PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 362 aa.	1.40E-196	
133	cg3001696	1154	CGCTGCACCTGT GCATCGCGCTGG G[C/T]TAGGCCAAT AGCAGCCTCAAC CCCCG	C	T	Gly	Gly	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa.	2.10E-195	1 (1p36.1)

134	cg3001696	815	TGGCTGTGACCC GTCCCGGGACG G[G/T]GCAGTGGT GTGCATGCTCCA GTTCC	G	T	Gly	Gly	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa.	2.10E-195	1 (1p36.1)
135	cg42704646	407	TGGCCTTCCCGAT CACCATGCTGCT C/G]ACTGGTTTCG TGGGCAACGCAC TGG	C	G	Leu	Leu	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.	3.10E-194	1 (1p31.2)
136	cg43326635	347	GGGATGCCACCT TCTGCTTCATCGT C/G]TCGCTGGCG GTGGCTGATGTG GCCC	C	G	Val	Val	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	1.10E-173	1
137	cg3003708	358	CCATCTCCTTCTG TGGCTGTCTCAC A/G]CAGATGTATT TCGTTTTCATGTT CG	A	G	Thr	Thr	SILENT- CODING	tm7	Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	2.50E-160	
138	cg3003708	787	GGTGAAAGCCT TCTCCACCTGTGG [T/C]TCTCACCTGG CTGTGGTTCTCCT CT	T	C	Gly	Gly	SILENT- CODING	tm7	Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	2.50E-160	



139	cg3003708	841	ACAGCACCATCAT TGCTGTGTAATTTT /C/JAACCCCTCTGTC CTCCCACTCAGCT G	T	C	Phe	Phe	SILENT- CODING	tm7	Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	2.50E-160	
140	cg36729339	537	ACTCTCCAATGTA CTTTTCCTCTC[C /T/JAACCTCTCCTT CTTGACCTCTGC T	C	T	Ser	Ser	SILENT- CODING	tm7	Human Gene SWISSPROT- ID:Q15062 OLFACTORY RECEPTOR-LIKE PROTEIN FAT11 - HOMO SAPIENS (HUMAN), 316 aa.	1.90E-153	
141	cg38841806	717	GACATCAGGCGC ACGGTGCCCAACC T[C/G]CGCCATCT GCAGGCCCAAGAA GAAGT	C	G	Leu	Leu	SILENT- CODING	tm7	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	2.10E-67	
142	cg38841806	723	AGCGGCACGGTG CCAACCTCCGCC AIT/CJCTGCAGGC CAAGAAGAAGTTT GTGA	T	C	His	His	SILENT- CODING	tm7	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	2.10E-67	
143	cg38841806	96	CAGCCTTCTCCAT GCCAGCTGGCA[ G/A]CTGGCACTGT GGGCACCAGCCT ACC	G	A	Gln	Gln	SILENT- CODING	tm7	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	2.10E-67	

144	cg43040273	1966	CCTGTGCTGATCT GGTCATGGGCCTI G/AJGCAGTGGTG CCCTTTGGGGCC GCC	G	A	Leu	Leu	SILENT- CODING	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
145	cg43040273	2237	CTTGCCCATTCAG ATGCACTGGTACI C/AJGGGCCACCC ACCAGGAAGCCA TCAA	C	A	Arg	Arg	SILENT- CODING	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
146	cg43336100	687	TTGGAAGCGTGC ATCCAGTGAGACC IATJATGAGGCTTG AGTCTTTTAGTGC CT	A	T	Pro	Pro	SILENT- CODING	tnf	Human Gene SWISSPROT- ID:P26022 PENTAXIN-RELATED PROTEIN PTX3 PRECURSOR (TUMOR NECROSIS FACTOR- INDUCIBLE PROTEIN TSG-14) - HOMO SAPIENS (HUMAN), 381 aa.	2.20E-207	3 (3q25)
147	cg21646034	376	GTGTGAGCAGAG ATGCCAGAACCAG IAGJGTGGACCGA ACACCATACATA TGG	A	G	Lys	Lys	SILENT- CODING	transcriptfac tor	Human Gene SWISSPROT- ID:Q06545 GA BINDING PROTEIN BETA-2 CHAIN (GABP-BETA-2 SUBUNIT) (TRANSCRIPTION FACTOR E4TF1-47) (GAPBP2) - HOMO SAPIENS (HUMAN), 347 aa.	9.00E-179	15

148	cg43916882	1608	TGGCAGCTACCA GCACACTGCCTC CJA/GJCCGTCAAT AAAGGCACTGATG GTCT	A	G	Gly	Gly	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P39656 DOLICHYL- DIPHOSPHOOLIGOSACCHARIDE-- PROTEIN GLYCOSYLTRANSFERASE 48 KD SUBUNIT PRECURSOR (EC 2.4.1.119) (OLIGOSACCHARYL TRANSFERASE 48 KD SUBUNIT) (DOST 48 KD SUBUNIT) (KIAA0115) (HA0643) - HOMO SAPIENS (HUMAN), 456 aa.	5.30E-245	1
149	cg2537639	294	TGGCTCCCATTTGT CTGGGAGGGCACI A/GITTCACATCG ACATCCTCAACGA GC	A	G	Thr	Thr	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P18442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYLTRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)

150	cg2537639	654	ACGTGGACATGG AGTTCCCGCACC A/C/TGTGGGCGT GGAGATCCTGACT CCGC	T	His	His	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYLTRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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151	cg2537639	678	ACGTGGGCGTGG AGATCCTGACTCC [G/A]CTGTTCGGC ACCCTGCACCCC GGCT	A	Pro	Pro	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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152	cg2537639	768	GGCCCCAGTCCC AGGCCCTACATCCC [C/T]AAGGACGAG GGCGATTCTACT ACC	T	Pro	Pro	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYLTRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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153	cg2537639	927	ACGAGAGCCACC TGAACAAGTACCT [G/A]CTGCGGCAC AAACCCACCAAG GTGC	G	A	Leu	Leu	SILENT- CODING	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS- FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
154	cg44000740	732	GGGAGATACTG GCTCACCCAGGA A[A/C]ACAGGGAA CATCACCTTATGC CACA	A	C	Val	Val	SILENT- CODING	transferase	Human Gene Homologous to SWISSPROT-ID:P30711 GLUTATHIONE S-TRANSFERASE THETA 1 (EC 2.5.1.18) (CLASS- THETA) - HOMO SAPIENS (HUMAN), 239 aa.	1.60E-117	16

155	cg38869466	1185	ACGCAGTGGCCG TGGGCTCCCTCT G[C]TGCTCTTTCC GCCAGTCTTCTAG GTT	C	T	Cys	Cys	SILENT- CODING	transport	Human Gene SWISSPROT- ID:P30825 HIGH-AFFINITY CATIONIC AMINO ACID TRANSPORTER-1 (CAT-1) (CAT1) (SYSTEM Y+ BASIC AMINO ACID TRANSPORTER) (ECOTROPIC RETROVIRAL LEUKEMIA RECEPTOR HOMOLOG) (ERR) (ECOTROPIC RETROVIRUS RECEPTOR HOMOLOG) - HOMO SAPIENS (HUMAN), 629 aa.	0	13
156	cg40351913	1347	CCATCGCCACGC TCCCTCTGTCTC [A/G]GCCTGGGCC GTGGTCTTCTTCA TCA	A	G	Ser	Ser	SILENT- CODING	transport	Human Gene SWISSPROT- ID:Q01959 SODIUM-DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	0	5 (5p15.3)
157	cg43964039	1719	GATGGAACAGCT CCTCGGGTGTCTT [G/A]TCACTTTGGC TGGCTCCCCCT GCC	G	A	Asp	Asp	SILENT- CODING	transport	Human Gene SWISSPROT- ID:P11166 GLUCOSE TRANSPORTER TYPE 1, ERYTHROCYTE/BRAIN - HOMO SAPIENS (HUMAN), 492 aa.	1.60E-259	1
158	cg43992017	1656	GCGGCTGCTGGT GGATGGGTGGGC G[C/G]GGGGTGCA GCCTCCACCCCC TCCCC	C	G	Pro	Pro	SILENT- CODING	transport	Human Gene SPTREMBL- ID:Q14728 TETRACYCLINE TRANSPORTER-LIKE PROTEIN MRNA - HOMO SAPIENS (HUMAN), 455 aa.	4.40E-241	



159	cg43948629	1238	CGCCTGTAATGG CTGTGAACATGCT [C/T]ACCCAGCAG GAGGTCCTGTC GTTA	C	T	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q15031 PROBABLE LEUCYL- TRNA SYNTHETASE, MITOCHONDRIAL PRECURSOR (EC 6.1.1.4) (LEUCINE--TRNA LIGASE) (LEURS) (KIAA0028) - Homo sapiens (Human), 903 aa.	0	3
160	cg43955093	2875	CATTGACTAGGG GCTGTGGGGCA T[C/G]CGCCAGG TGCCCTCCATCA GAGG	C	G	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	0	16
161	cg43955093	3385	AGCAGGCCAAGA GAGATCTGTGAA [C/T]GCATCTTGT CCAGAATACCAGA TA	C	T	Ala	Ala	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	0	16
162	cg43055918	517	CGCTGGCATAGG ACATGGCGGGCT T[G/T]CCCCCGC AGAGCTCTGGGG GCTAC	G	T	Gly	Gly	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	0	17
163	cg43974592	254	AAATAACAAGGCA TTGAAGAAATGGC[ T/A]GACGAGCGG AAAGACGAAGGA AAG	T	A	Ala	Ala	SILENT- CODING	UNCLASSI FIED	Human Gene REMTREMBL- ACC:E1296438 SEQUENCE 28 FROM PATENT WO9727323 - UNIDENTIFIED, 1829 aa.	0	2 (2q34)

164	cg43956384	206	AAGGACGCAACG CTGCCACCATGG A[C/T]AGTAGCAC CTGGAGCCCCAA GACCA	C	T	Asp	Asp	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P13866 SODIUM/GLUCOSE COTRANSPORTER 1 (NA+)/GLUCOSE COTRANSPORTER 1) (HIGH AFFINITY SODIUM-GLUCOSE COTRANSPORTER) - Homo sapiens (Human), 664 aa.	0	22 (22q13.1 )
165	cg44025634	2757	TGAAAGTATTCAA TCCCAGAAGGAA A/G]CTGGAATTG CCCTTCTGTTCT AG	A	G	Lys	Lys	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSNEW- ACC:P00450 CERULOPLASMIN PRECURSOR (EC 1.16.3.1) (FERROXIDASE) - Homo sapiens (Human), 1065 aa.	0	3 (3q21)
166	cg43940037	2472	GCTGGCGCACCTG CTAGCCTCAGAG G[T/A]GCCAGCAC CTCCTCAGCCCC CGCGC	T	A	Ala	Ala	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P41250 GLYCYL-TRNA SYNTHETASE (EC 6.1.1.14) (GLYCINE--TRNA LIGASE) (GLYRS) - Homo sapiens (Human), 685 aa.	0	7 (7p15)
167	cg44024279	481	AAACCAGCTACC TGCCTTTCTGGA A/G]GAACCTTTGCC ATGAGAAAGAAAT TT	A	G	Glu	Glu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P02771 ALPHA- FETOPROTEIN PRECURSOR (ALPHA-FETOGLOBULIN) (ALPHA- 1-FETOPROTEIN) - Homo sapiens (Human), 609 aa.	0	

168	cg43926814	1122	CATGAGTTTGGAT CCAGCTCTTCTI C/TCCCTGGCTT TCTGGGCCATTTG TC	C	T	Glu	Glu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q13573 NUCLEAR PROTEIN SKIP (SNW1 PROTEIN) (NUCLEAR RECEPTOR COACTIVATOR NCOA- 62) - Homo sapiens (Human), 536 aa.	5.00E-289	14
169	cg40918088	1778	TTGGAGCTGGAAT TACTGTGTATGA[A /G]GCCTTAGCAGC TGCTGATGAGCTT T	A	G	Glu	Glu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P51854 TRANSKETOLASE 2 (EC 2.2.1.1) (TK 2) (TRANSKETOLASE RELATED PROTEIN) - Homo sapiens (Human), 557 aa.	1.80E-287	X (Xq28)
170	cg43966985	1242	TCAACACCTACGT CCACTTCCAAGG[ G/T]AAGATGAAGG GCTTCTCCCTGCT GG	G	T	Gly	Gly	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P01019 ANGIOTENSINOGEN PRECURSOR - Homo sapiens (Human), 485 aa.	3.90E-257	1 (1q42)
171	cg43924009	770	TGGCTTGCACAA TTGCTTGAAGAC[ A/T]CGATCCATGT AAGTGGAAGTGTCT TG	A	T	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q43411 HYPOTHETICAL 49.3 KD PROTEIN - HOMO SAPIENS (HUMAN), 442 aa (fragment).	6.90E-239	2 (2cen)
172	cg42913861	2186	CTGGGCAGCTGC CCTCACAGTAGTT [C/G]CCGTAAGTAG CCGGTGGGTGCT ATGA	C	G	Gly	Gly	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.	3.00E-227	2 (2cen)

173	cg42913861	2354	GCCGAGCCTGCA CCACCACAAAGG G/C/T/CGGTGCGA CTCTTCGCCTGG GTCCA	C	T	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.	3.00E-227	2 (2cen)
174	cg43929685	256	CATAGAAAGGCCA GGAGTCAGGAGA C/C/T/TGGGTTCT GTCCTGGATTATA CACC	C	T	Gln	Gln	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P29080 (2'- 5')OLIGOADENYLATE SYNTHETASE 1B (EC 2.7.7.-) (2- 5')OLIGO(A) SYNTHETASE 1B) (2- 5A SYNTHETASE 1B) - Mus musculus (Mouse), 414 aa.	2.40E-225	12
175	cg43929685	268	GGAGTCAGGAGA CCTGGGTTCTGTC [C/T]TGGATTATAC ACCAGCTCACTGA GG	C	T	Gln	Gln	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P29080 (2'- 5')OLIGOADENYLATE SYNTHETASE 1B (EC 2.7.7.-) (2- 5')OLIGO(A) SYNTHETASE 1B) (2- 5A SYNTHETASE 1B) - Mus musculus (Mouse), 414 aa.	2.40E-225	12
176	cg43918561	53	CCATGCCACCCC CCGACGCCACCA C/G/C/CACAGGC CAAGGGCTTCCG CAGGG	G	C	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P04177 TYROSINE 3- MONOOXYGENASE (EC 1.14.16.2) (TYROSINE 3-HYDROXYLASE) (TH) - Rattus norvegicus (Rat), 498 aa.	2.10E-224	11 (11p15.5)

177	cg42343176	1885	ATTTAATGAATTC CTGAAGACTGT GJAGAACTACAA TGAGAAATCCCTT T	A	G	Val	Val	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P14902 INDOLEAMINE 2,3- DIOXYGENASE (EC 1.13.11.42) (IDO) (INDOLEAMINE- PYRROLE 2,3-DIOXYGENASE) - Homo sapiens (Human), 403 aa.	3.90E-218	8 (8p12)
178	cg43956382	1146	AAAACAATGATAT CGATGAAGTTAT C/TJATCCACAG CTCCCTTATACAA AC	C	T	Ile	Ile	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q99816 TUMOR SUSCEPTIBILITY PROTEIN - HOMO SAPIENS (HUMAN), 390 aa.	4.90E-211	11
179	cg43984681	979	CACCATGAAGCA GTTGCTGCGGC C/C/TJTGAGGAG GCCGCGTGCGG GAAGT	C	T	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:O15382 BRANCHED-CHAIN AMINO ACID AMINOTRANSFERASE, MITOCHONDRIAL PRECURSOR (EC 2.6.1.42) (BCAT(M)) - Homo sapiens (Human), 392 aa.	1.30E-210	19 (19q13)
180	cg43984681	1074	TCCTGTACAAAGA CAGGAACCTCCA C/TJATCCACCA TGGAATGCGC CTG	C	T	His	His	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:O15382 BRANCHED-CHAIN AMINO ACID AMINOTRANSFERASE, MITOCHONDRIAL PRECURSOR (EC 2.6.1.42) (BCAT(M)) - Homo sapiens (Human), 392 aa.	1.30E-210	19 (19q13)

181	cg43950996	1762	CTGCGGTGGAGA CGTCAGAGCTGC C[A/G]GGGGAGGG GGCTCCTGCGCC ACAGC	A	G	Pro	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:P78545 ESE-1B - HOMO SAPIENS (HUMAN), 371 aa.	6.20E-204	1
182	cg44024506	988	ACCAGCTGCTCGT AGTACACAGGCA G/A]GCACTTCTCC TTGCCTACCTCCA TG	G	A	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:O60704 TYROSYLPROTEIN SULFOTRANSFERASE-2 - HOMO SAPIENS (HUMAN), 377 aa.	1.90E-200	22
183	cg43980381	1114	CTACCGCCAACTA TGACTTTGTCCTJC /G]AAGAAGCGGA CCTTCACCAAGG GAG	C	G	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q03385 GUANINE NUCLEOTIDE DISSOCIATION STIMULATOR RALGDS FORM A (RALGEF) - Mus musculus (Mouse), 852 aa.	5.60E-191	9



187	cg43248117	1054	AACCAAGCCCACT GTGAGAAGACCA C/G/C/GTGTTCAA GTC TTGGGAATG GCAG	G	C	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSPROT- ACC:Q14894 MU-CRYSTALLIN HOMOLOG (NADP-REGULATED THYROID-HORMONE BINDING PROTEIN) - Homo sapiens (Human), 314 aa.	1.20E-161	16 (16p13.1 1)
188	cg44027049	482	CCACAAATGTTAGG AGGGTATTTTAAIC /TATCCCTCCAGT TAACAAATACAGC A	C	T	Tyr	Tyr	SILENT- CODING	UNCLASSI FIED	Human Gene SWISSNEW- ACC:P11245 ARYLAMINE N- ACETYLTRANSFERASE, POLYMORPHIC (EC 2.3.1.5) (PNAT) (NAT-2) (ARYLAMINE ACETYLASE) - Homo sapiens (Human), 290 aa.	5.40E-157	8 (8p23.1)
189	cg43982075	499	CTGCCATCTTTCA GCCCTCTGAAACI C/TJGTGCCAGCA CAGAACTCTCCCT GG	C	T	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q15729 THYROTROPH EMBRYONIC FACTOR - HOMO SAPIENS (HUMAN), 303 aa.	1.20E-154	22
190	cg43942977	350	GCGCTTCCCAGG TCCGGACAATTCG [G/T]CAGACTATTG TCAAACTGGGAA TA	G	T	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	9.60E-148	
191	cg43942977	701	GGCAGCTGAAGA TCACCAATGCTGG [G/C]ATGGTGCTCT GATGAGGAGTTG GAGC	G	C	Gly	Gly	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	9.60E-148	



192	cg43942977	773	GCGAGGTGTTGT GTCCAATATCCTT G/TAAAGGACACGC AGGTGACTCGAC AGG	G	T	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	9.60E-148	
193	cg43985220	753	TGTACACTGCCAG AAAAGGAAAAGG T/GGCCCTTTGTA ATGGTCAAAAAC AC	T	G	Gly	Gly	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:P29218 MYO- INOSITOL-1(OR 4)- MONOPHOSPHATASE (EC 3.1.3.25) (IMP) (INOSITOL MONOPHOSPHATASE) (LITHIUM- SENSITIVE MYO-INOSITOL MONOPHOSPHATASE A1) - Homo sapiens (Human), 277 aa.	5.10E-145	8
194	cg43985220	837	TCTTGGTGACTGA GTTGGGCTCTTC C/TAGAACACCCAG AGACTGTGAGAA GG	C	T	Ser	Ser	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:P29218 MYO- INOSITOL-1(OR 4)- MONOPHOSPHATASE (EC 3.1.3.25) (IMP) (INOSITOL MONOPHOSPHATASE) (LITHIUM- SENSITIVE MYO-INOSITOL MONOPHOSPHATASE A1) - Homo sapiens (Human), 277 aa.	5.10E-145	8
195	cg43946394	321	TAGAGCGCACAC AGGCCTCCAGCT G/A/GGCCCATGTC CGTCTCATCATCC CAAG	A	G	Ala	Ala	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P29692 ELONGATION FACTOR 1-DELTA (EF-1-DELTA) - Homo sapiens (Human), 281 aa.	2.80E-144	19

196	cg43119818	1329	CTGACAGCTACAG GCTCTTTTCAGTTTC TTCATTTTCACTG GGGCAGTACAAAT G	C	T	Phe	Phe	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P00915 CARBONIC ANHYDRASE I (EC 4.2.1.1) (CARBONATE DEHYDRATASE I) - Homo sapiens (Human), 260 aa.	6.90E-141	8 (8q22)
197	cg43118279	735	AGAAAGTTGAAGG GGCTGGTGCCAC TTT/GGGACCCGA ATCAAGTCGACAC ACTA	T	G	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:Q05195 MAD PROTEIN (MAX DIMERIZER) - Homo sapiens (Human), 221 aa.	1.20E-111	2 (2p13)
198	cg43325007	866	TGGGTTTCAGGGA TGAGCCCTTCTC TTT/CJACAGCCAGG CGGCTCAGGGCA AACA	T	C	Val	Val	SILENT- CODING	UNCLASSI FIED	Human Gene Homologous to TREMBLNEW-ACC:AAD43195 PEROXISOMAL MEMBRANE PROTEIN PMP 24 - HOMO SAPIENS (HUMAN), 212 aa.	4.80E-110	20
199	cg39524111	402	GCCAAATATAGGAT AGGGCACTACAGT A/GJTCCGGTACA GTGACACCCCTGG AGC	A	G	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:BAA13472 CD89_U08 - HOMO SAPIENS (HUMAN), 191 aa.	2.10E-100	19 (19q13.4)
200	cg43280516	629	ACGGGGAGGAGC TGCAGATGGAAC C/C/TJGTGTGAGG TGCTTCTGGGAC CTGC	C	T	Pro	Pro	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:CAB43107 PRENYLATED RAB ACCEPTOR 1 (PRA1) - HOMO SAPIENS (HUMAN), 185 aa.	6.80E-95	19

201	cg43963913	871	AGAGGTTGGGGG GCGCCGAGCGCG A[G/A]CGGCCCG AAAGGGGCTGGG CTCCT	G	A	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O14803 BCL-X/BCL-2 BINDING PROTEIN - HOMO SAPIENS (HUMAN), 168 aa (fragment).	5.10E-90	11
202	cg40262905	682	TAGTAAAGGCCT GAAATATATGCTTG /C/GAGGTGGAAT TGGCAGAACTACC T	G	C	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:BAA34941 HUMAN CMAP - HOMO SAPIENS (HUMAN), 167 aa.	1.30E-89	
203	cg43918168	915	CTCCATCAACAGC ATCCGGACTGCA[ T/C]GGCGGCTCG CCGTGCGGCTGG GGCC	T	C	Pro	Pro	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P09496 CLATHRIN LIGHT CHAIN A (BRAIN AND LYMPHOCYTE LCA) - Homo sapiens (Human), 248 aa.	3.80E-85	9 (12q23)
204	cg43259701	136	CGACGAGGTGCT ACGCGAGGGCGA G[C/T]TGAGAAG CGCAGCGACAGC CTCTT	C	T	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O00496 IPL (IPL) - HOMO SAPIENS (HUMAN), 152 aa.	1.30E-77	11
205	cg1527767	162	TTTTTCCAGCTT ACAAATGGTACAG[ A/G]CAGGAGCCT GGGGAAGGTCCT GTCC	A	G	Arg	Arg	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G36907 T-CELL RECEPTOR ALPHA-CHAIN HAP58 V(A)10.1-J(A)T - HOMO SAPIENS (HUMAN), 135 aa (fragment).	5.60E-68	

206	cg40968986	316	AGAAGAGAGCCT GTGACACTGCCA C/C/T/TGTGTGACT CATCGGCTGGCA GGCT	C	T	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P06881 CALCITONIN GENE- RELATED PEPTIDE I PRECURSOR (CGRP-I) (ALPHA-TYPE CGRP) - Homo sapiens (Human), 128 aa.	5.10E-58	11 (11p15.2 )
207	cg42550133	300	TCATCCTGAGTTC TAAGAAAGCTCCTT /C/CTCAGTGACTC TGGCTTCATCTC T	T	C	Leu	Leu	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:D1002898 T- CELL RECEPTOR BETA-CHAIN V REGION - HOMO SAPIENS (HUMAN), 112 aa (fragment).	8.50E-56	7 (7q35)
208	cg2526759	317	CTCTGGTTGTCCA CGAGGGAGACACI T/C]GTAACCTCA ATTGCAGTTATGA AG	T	C	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	
209	cg41664708	249	AAGTCGTGCTGA TCCACAAGCCACI A/G]TGCGTGAGA GACGTGGTCAGG AGCA	A	G	Thr	Thr	SILENT- CODING	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P47992 LYMPHOTACTIN PRECURSOR (CYTOKINE SCM-1) (ATAC) (LYMPHOTAXIN) (SCM-1- ALPHA) - Homo sapiens (Human), 114 aa.	2.00E-54	1
210	cg43300673	1571	AGGGAGGCGGGG AGGGTAGCATGG G[G/gap]CACACGG CCCTCACAGGGA CTCACT	G	gap			SILENT- NONCODI NG	ATPase_as sociated	Human Gene SPTREMBL- ID:Q93050 VACUOLAR-TYPE H(+)- ATPASE 115 KDA SUBUNIT - HOMO SAPIENS (HUMAN), 831 aa.	0	17

211	cg43284434	2570	AGTTGAAATCAGA GAGGAATAAAAA[ ap/A]GACATTTTAT ATTTTATTCTGCT CC	gap	A				SILENT- NONCODI NG	ATPase_as sociated	Human Gene Homologous to SPTREMBL-ID:Q18788 C52E4.5 - CAENORHABDITIS ELEGANS, 590 aa.	4.00E-121	6
212	cg43132502	196	TAAGCATGAGGTG GCACGAGGCAGG[ A/C]GTTGGCGATG CCACCTGGGGGT CAC	A	C				SILENT- NONCODI NG	ATPase_as sociated	Human Gene Similar to SPTREMBL- ID:Q15332 GAMMA SUBUNIT OF SODIUM POTASSIUM ATPASE LIKE - HOMO SAPIENS (HUMAN), 126 aa.	9.40E-58	11
213	cg43931765	606	GGTCCCCTTGCTT TATCCCAAGCTC[ G/T]GAGGGACGC AGCCTGGCATGG CTCT	G	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
214	cg43931765	607	GTCCCCTTGCTT ATCCCAAGCTCG[ G/T]AGGGACGCA GCCTGGCATGGC TCTG	G	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
215	cg43931765	615	CTTTATCCCAAGC TCGGAGGGACGC[ gap/G]AGCCTGGC ATGGCTCTGGCCT AGCA	gap	G				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3

216	cg43931765	660	TAGCAGCCAGGT GACATGGCCAGG C[gap]/TJACCTCC TGACAGGCACTG TGGGC	gap	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
217	cg43931765	665	GCCAGGTGACAT GGCCAGGCACCT T[gap]/TJCCTGTAC AGGCACTGTGG CTCCTG	gap	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
218	cg43931765	668	AGGTGACATGGC CAGGCACCTTCCT [gap]/TGTACAGGC ACTGTGGGCTCCT GGCC	gap	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
219	cg43931765	668	AGGTGACATGGC CAGGCACCTTCCT [gap]/TGTACAGGC ACTGTGGGCTCCT GGCC	gap	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3
220	cg43931765	668	AGGTGACATGGC CAGGCACCTTCCT [gap]/TGTACAGGC ACTGTGGGCTCCT GGCC	gap	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	0	3

221	cg43952088	4769	AATCCACAATCGG CATCAGGAAGCCI A/CJAAAGTCCCAGT GGCCATTAGGGT CCT	A	C				SILENT- NONCODI NG	cadherin	Human Gene SPTREMBL- ID:Q15065 OB-CADHERIN-1 - HOMO SAPIENS (HUMAN), 796 aa.	0	16
222	cg44010957	1406	TCCCTATGAGCCT GCAAAGGAGACA[ G/T]TCAGGAATGA GTTCCATGTTCTGA GA	G	T				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P20701 LEUKOCYTE ADHESION GLYCOPROTEIN LFA-1 ALPHA CHAIN PRECURSOR (LEUKOCYTE FUNCTION ASSOCIATED MOLECULE 1, ALPHA CHAIN) (CD11A) (INTEGRIN ALPHA-L) - HOMO SAPIENS (HUMAN), 1170 aa.	0	16 (16p11.2 )
223	cg43956560	1463	CAGTGCATCTGG GAAGATTTCCTACCI T/CJGACCAACAGT TCCTTCAGCTTCC AT	T	C				SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90-MEL) (LEUKOCYTE-ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAM1) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	1.00E-218	1 (1q23)

224	cg43956560	1492	CAACAGTTCCTTC AGCTTCCATTTCIG /AJCCCTCATTTA TCCCTCAACCCCC A	G	A			SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90-MEL) (LEUKOCYTE-ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAM1) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	1.00E-218	1 (1q23)
225	cg43956560	2242	TGCTCTCCTTTCC CCTGCCCCACAG C/AJCTTTATCCA CTTACCCTAGATTC TA	C	A			SILENT- NONCODI NG	cadherin	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90-MEL) (LEUKOCYTE-ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAM1) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	1.00E-218	1 (1q23)



226	cg43264626	428	TGGCCACAGTGA AAAAGGTCATGG G[T/A]GGAGAGAA GCAAAAGTAGGAA GGATC	T	A				SILENT- NONCODI NG	cathepsin	Human Gene SWISSPROT- ID:P43235 CATHEPSIN K PRECURSOR (EC 3.4.22.38) (CATHEPSIN O) (CATHEPSIN X) (CATHEPSIN O2) - HOMO SAPIENS (HUMAN), 329 aa.	4.10E-183	1
227	cg43011543	1972	ACCGCACCCCTTTC CACCCGGTGGGG [C/G]CCCAGTGAA GTTTAACAAACTG CTG	C	G				SILENT- NONCODI NG	collagen	Human Gene SWISSPROT- ID:P27658 COLLAGEN ALPHA 1(VIII) CHAIN PRECURSOR (ENDOTHELIAL COLLAGEN) - HOMO SAPIENS (HUMAN), 744 aa.	0	
228	cg43011543	2096	CATACCACGTTCA CTGCAAGGGGGG[ C/G]AACGTGTGG GTTGCTCTATTCA AGA	C	G				SILENT- NONCODI NG	collagen	Human Gene SWISSPROT- ID:P27658 COLLAGEN ALPHA 1(VIII) CHAIN PRECURSOR (ENDOTHELIAL COLLAGEN) - HOMO SAPIENS (HUMAN), 744 aa.	0	
229	cg43933757	2546	GAACCCAGTAG GCTCCTGGAGGC C[A/C]TGGTCAGC TTGCTTGAATCC AGCA	A	C				SILENT- NONCODI NG	complement	Human Gene SWISSPROT- ID:P10643 COMPLEMENT COMPONENT C7 PRECURSOR - HOMO SAPIENS (HUMAN), 843 aa.	0	5 (5p13)
230	cg41553795	64	TGGTGGTGCTAC CCTTGGCCTCCCA [C/G]AGTCCTGCC ACCCTGCTGCCG CCAC	C	G				SILENT- NONCODI NG	complement	Human Gene Homologous to SWISSPROT-ID:P07360 COMPLEMENT C8 GAMMA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 202 aa.	1.40E-104	9 (9q34.3)

231	cg42542496	168	AGCCCTTCTCCAC CCGGATAGATTCT CTTTCACCCCTTGG CCCGCCTTTGCC CCA	C	T				SILENT- NONCODI NG	csf	Human Gene SWISSPROT- ID:P40225 THROMBOPOIETIN PRECURSOR (MEGAKARYOCYTE COLONY STIMULATING FACTOR) (C-MPL LIGAND) (ML) (MEGAKARYOCYTE GROWTH AND DEVELOPMENT FACTOR) (MGDF) - HOMO SAPIENS (HUMAN), 353 aa.	1.20E-189	3 (3q26.3)
232	cg42542496	179	ACCCGGATAGATT CCTCACCCCTTGGI CTJCCGCCCTTTGC CCACCCCTACTCT GC	C	T				SILENT- NONCODI NG	csf	Human Gene SWISSPROT- ID:P40225 THROMBOPOIETIN PRECURSOR (MEGAKARYOCYTE COLONY STIMULATING FACTOR) (C-MPL LIGAND) (ML) (MEGAKARYOCYTE GROWTH AND DEVELOPMENT FACTOR) (MGDF) - HOMO SAPIENS (HUMAN), 353 aa.	1.20E-189	3 (3q26.3)
233	cg41533258	1356	GTGCTGGACATT TGCCCTTGCTGGA CTJGGGGACTGG GGATGTGGGAGG GAGC	C	T				SILENT- NONCODI NG	csf	Human Gene Homologous to SWISSPROT-ID:P09919 GRANULOCYTE COLONY- STIMULATING FACTOR PRECURSOR (G-CSF) (PLURIPROETIN) - HOMO SAPIENS (HUMAN), 207 aa.	1.50E-107	17 (17q11.2)

234	cg2753430	657	ACGACTTTGAGCC TCGCGATCTTTTIG ap/GJAGTCCAACG TCCAGCTCGTTCT CTG	G			SILENT- NONCODI NG	csf	Human Gene Similar to SWISSNEW- ID:P08700 INTERLEUKIN-3 PRECURSOR (IL-3) (MULTIPOTENTIAL COLONY- STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (P-CELL STIMULATING FACTOR) (MAST-CELL GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HUMAN), 152 aa.lpcis:SWISSPROT-ID:P08700 INTERLEUKIN-3 PRECURSOR (IL- 3) (MULTIPOTENTIAL COLONY- STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (P-CELL STIMULATING FACTOR) (MAST-CELL GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HUMAN), 152 aa.	1.10E-77	5
235	cg44036323	225	TGCGGCTTAAAG GGCAACCCCGCGC G/CJGGACCCCTTCC TCCCTAGTCGCG GGG	G	C		SILENT- NONCODI NG	dehydrogen ase	Human Gene SWISSPROT- ID:P00367 GLUTAMATE DEHYDROGENASE 1 PRECURSOR (EC 1.4.1.3) (GDH) - HOMO SAPIENS (HUMAN), 558 aa.	5.80E-303	10

236	cg43918671	766	GAGAGACCATTTA CTTACATCAGTT[C /T]GGTTTATAGAC ATTTGAATCATAT C	C	T				SILENT- NONCODI NG	dehydrogen ase	Human Gene SPTREMBL- ID:Q14131 DIHYDROLIPOAMIDE DEHYDROGENASE - HOMO SAPIENS (HUMAN), 511 aa.	5.10E-272	7 (7q31)
237	cg43057018	1995	AGTTTCATTATAC TTTTCTCTCCAC[g ap/G]TTTTGTCTAT GTTGAAAAATTTTC TG	gap	G				SILENT- NONCODI NG	dehydrogen ase	Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.  pcis:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.	1.30E-209	4 (4q22)
238	cg44005808	3691	ACAAGACAGAAG CTGAAGTGCATCC [gap/C]AAAGGTGC TCAGAGAGCCGG CCCCG	gap	C				SILENT- NONCODI NG	dna_ma_bi nd	Human Gene SWISSNEW- ID:P19838 NUCLEAR FACTOR NF- KAPPA-B P105 SUBUNIT (DNA- BINDING FACTOR KBF1) (EBP- 1) [CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P50 SUBUNIT] - HOMO SAPIENS (HUMAN), 969 aa.  pcis:SWISSPROT-ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF- KAPPA-B P50 SUBUNIT) (DNA-BINDING FACTOR KBF1) (EBP-1) - HOMO SAPIENS (HUMAN), 969 aa.	0	

239	cg44005808	630	TCCTCCTCTCCA GCCGGCAGGCC [gap/G]CGCCGCTT AGGAGGGAGAGC CCACC	gap	G			SILENT- NONCODI NG	dna_rna_ nd	Human Gene SWISSNEW- ID:P19838 NUCLEAR FACTOR NF- KAPPA-B P105 SUBUNIT (DNA- BINDING FACTOR KBF1) (EBP- 1) [CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P50 SUBUNIT] - HOMO SAPIENS (HUMAN), 969 aa. lpcds:SWISSPROT-ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF- KAPPA-B P50 SUBUNIT) (DNA-BINDING FACTOR KBF1) (EBP-1) - HOMO SAPIENS (HUMAN), 969 aa.	0	
240	cg43956159	1244	TGGCGAGTCCAG GGTCACCCACATA [gap/A]CCATGCAC CACGGGTGCTAT GCCGC	gap	A			SILENT- NONCODI NG	dna_rna_ nd	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	1.40E-159	10
241	cg43956159	1248	GAGTCCAGGGTC ACCCACATACCAT [gap/T]GCACCACG GGTGCTATGCCG CTTCT	gap	T			SILENT- NONCODI NG	dna_rna_ nd	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	1.40E-159	10

242	cg43956159	1268	TACCATGCACCCAC GGGTGCTATGCC[ G/A]CTTCTTACAG GACCTTTTATAGCC CT	G	A				SILENT- NONCODING	dna_ma_bi nd	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	1.40E-159	10
243	cg43956159	1342	CCTGGAGGCAAC TGGGTAGGGTGC A[G/C]AACGGCAT GCTTTGGCTGGAA CACG	G	C				SILENT- NONCODING	dna_ma_bi nd	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	1.40E-159	10
244	cg43956159	1364	CAGAACGGCATG CTTTGGCTGGAAC [gap/C]ACGCATCC CTCCTTCCACGGC CGGC	gap	C				SILENT- NONCODING	dna_ma_bi nd	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	1.40E-159	10
245	cg43971258	471	CAGAGCTAGCTCT GGCTCTTCAGGC[ C/T]ACAAGTTAC AGTCCCTTCGCTCC TG	C	T				SILENT- NONCODING	dna_ma_bi nd_inhib	Human Gene Similar to SWISSNEW- ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX- LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa [pds:SWISSPROT-ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX-LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa.	1.30E-60	1 (1p36.13 )

246	cg43971258	508	GTCCTTCGCTCCT GAGCACCCAGGTI T/C/AGTCTCCAGG AAGGGATTGGTG AA	T	C				SILENT- NONCODI NG	dna_rna_bi nd_inhib	Human Gene Similar to SWISSNEW- ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX- LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa. Jpcls:SWISSPROT-ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX-LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa.	1.30E-60	1 (1p36.13 )
247	cg43982507	3373	GATACCTTTGCGT GGATCAAGCTTG gap/CJGTGACTTGA CCGTTTTATATTA CT	gap	C				SILENT- NONCODI NG	eph	Human Gene SWISSPROT- ID:P98155 VERY LOW-DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	0	9 (9p24)
248	cg43982507	3739	CAAAAAAATTTAT AAACTAATTTTGlg ap/GJTACGTATGA ATGATATCTTTGA CCT	gap	G				SILENT- NONCODI NG	eph	Human Gene SWISSPROT- ID:P98155 VERY LOW-DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	0	9 (9p24)
249	cg43982507	514	CCTCCTTCTCCCC CTTCCCCCTCCC A/C/GCCCCCACCT TCTTCCTCCTTTC GG	A	C				SILENT- NONCODI NG	eph	Human Gene SWISSPROT- ID:P98155 VERY LOW-DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	0	9 (9p24)

250	cg41554010	1371	CTGCCCTGCCAC CTGCTGTCTGTC [gap]/TCCAAAGAA GTTCTGGTATGAA CTTG	gap	T				SILENT- NONCODI NG	eph	Human Gene SWISSNEW- ID:P06727 APOLOPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.lpcis:SWISSPROT-ID:P06727 APOLOPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	1.80E-203	11 (11q23)
251	cg41554010	1371	CTGCCCTGCCAC CTGCTGTCTGTC [gap]/TCCAAAGAA GTTCTGGTATGAA CTTG	gap	T				SILENT- NONCODI NG	eph	Human Gene SWISSNEW- ID:P06727 APOLOPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.lpcis:SWISSPROT-ID:P06727 APOLOPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	1.80E-203	11 (11q23)
252	cg43984905	2376	TCCCTCCAGGACT AGGCTGGAGGAA[ G/C]CCAGTGGGG TCCCCCTGAGT GGGC	G	C				SILENT- NONCODI NG	esterase	Human Gene SWISSPROT- ID:P51178 1- PHOSPHATIDYLINOSITOL-4,5- BISPHOSPHATE PHOSPHODIESTERASE DELTA 1 (EC 3.1.4.11) (PLC-DELTA-1) (PHOSPHOLIPASE C-DELTA-1) (PLC-III) - HOMO SAPIENS (HUMAN), 756 aa.	0	3



253	cg43984905	2440	CACATGTGGGGA CAGGGCTGGTGT G G C TGTCTCCC AGCCTCTTGCTCA GAGC	G	C				SILENT- NONCODI NG	esterase	Human Gene SWISSPROT- ID:P51178 1- PHOSPHATIDYLINOSITOL-4,5- BISPHOSPHATE PHOSPHODIESTERASE DELTA 1 (EC 3.1.4.11) (PLC-DELTA-1) (PHOSPHOLIPASE C-DELTA-1) (PLC-III) - HOMO SAPIENS (HUMAN), 756 aa.	0	3
254	cg43992911	382	CTAAAGTCGGAGT ATCTTCTTCCAA G /A ATTTACAGTCT TGCGGCCCGTTT CA	G	A				SILENT- NONCODI NG	glycoprotein	Human Gene SWISSPROT- ID:P08183 MULTIDRUG RESISTANCE PROTEIN 1 (P- GLYCOPROTEIN 1) - HOMO SAPIENS (HUMAN), 1280 aa.	0	7
255	cg43932434	267	TTTCTAGAGGGG GTCTGTTGAAGAT  G A TGTAAGTAGT ACACCCCAACCC CCA	G	A				SILENT- NONCODI NG	glycoprotein	Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HEPARAN SULFATE PROTEOGLYCAN) (EPICAN) (CDW44) - HOMO SAPIENS (HUMAN), 742 aa.	1.80E-195 11 (11pter)	

256	cg43932434	306	CCCCAACCCCA ACCTCAGTGGAAA  A/G CAATGCCCA GGGATTAGGCTAT GGA	A	G				SILENT- NONCODI NG	glycoprotein	Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HEPARAN SULFATE PROTEOGLYCAN) (EPICAN) (CDW44) - HOMO SAPIENS (HUMAN), 742 aa.	1.80E-195	11 (11pter)
257	cg43318219	366	GCGCAGGTCAGA GGCGGCCGCGCAG C A/G GGCCTCCG CGAGGTCCCCAC GCCGG	A	G				SILENT- NONCODI NG	glycoprotein	Human Gene SWISSNEW- ID:P15813 T-CELL SURFACE GLYCOPROTEIN CD1D PRECURSOR (CD1D ANTIGEN) (R3G1) - HOMO SAPIENS (HUMAN), 335 aa. pcis:SWISSPROT-ID:P15813 T- CELL SURFACE GLYCOPROTEIN CD1D PRECURSOR (CD1D ANTIGEN) (R3G1) - HOMO SAPIENS (HUMAN), 335 aa.	3.10E-185	1 (1q21)

258	cg43967861	1954	CTCTATACTGTAC ACTCACCCATAA[T /gap]TCAAACAATT ACACCATGGTATA AA	T	gap			SILENT- NONCODI NG	glycoprotein	Human Gene Similar to SWISSPROT-ID:Q08878 FIBULIN-1, ISOFORM C PRECURSOR (BASEMENT-MEMBRANE PROTEIN 90) (BM-90) - MUS MUSCULUS (MOUSE), 685 aa.	8.20E-67	2
259	cg43967861	1955	TCTATACTGTACA CTCACCCATAA[T /gap]CAAACAATT CACCATGGTATAA AG	T	gap			SILENT- NONCODI NG	glycoprotein	Human Gene Similar to SWISSPROT-ID:Q08878 FIBULIN-1, ISOFORM C PRECURSOR (BASEMENT-MEMBRANE PROTEIN 90) (BM-90) - MUS MUSCULUS (MOUSE), 685 aa.	8.20E-67	2
260	cg43965366	1411	GCCGAATAGCCT GGGTTTGGAAA G[C/T]ATGTTTTG AAATATGTGGAT CTC	C	T			SILENT- NONCODI NG	glycoprotein	Human Gene Similar to SWISSPROT-ID:P49222 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - MUS MUSCULUS (MOUSE), 690 aa.	8.90E-61	6 (6p25)
261	cg43965366	385	TACTGACCCTAAAT CACACCCTAGAC[ A/T]TATCAGAGGG AAATTCTGACCAT AA	A	T			SILENT- NONCODI NG	glycoprotein	Human Gene Similar to SWISSPROT-ID:P49222 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - MUS MUSCULUS (MOUSE), 690 aa.	8.90E-61	6 (6p25)

262	cg43322513	1255	TGTCCTTGAAGAA CATGCACTTGGCI A/GICGGATGGCA CAAGCAAAATGGT AGA	A	G				SILENT- NONCODI NG	glycoprotein	Human Gene Similar to SWISSPROT-ID:P13983 EXTENSIN PRECURSOR (CELL WALL HYDROXYPROLINE-RICH GLYCOPROTEIN) - NICOTIANA TABACUM (COMMON TOBACCO), 620 aa.	3.30E-54	12
263	cg41637704	1397	CCCGCGCCCCAG TAGGAGCCCCGC G/gap/GjCCCAGCA GGTGGCGCGCGC ACGGAG	gap	G				SILENT- NONCODI NG	homeobox	Human Gene SWISSPROT- ID:P50219 HOMEBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	1.20E-224	7
264	cg41637704	1423	CCAGCAGGTGCG GCGCGCACGGAG C/gap/GjCGCCGG CCGGCGGCTTCT CCCGGAG	gap	G				SILENT- NONCODI NG	homeobox	Human Gene SWISSPROT- ID:P50219 HOMEBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	1.20E-224	7
265	cg41637704	1817	TGAAACTTGAAAC CGCCTCTGGAGCI C/TjGCCATTCTGC AGAGTATTGGAA AA	C	T				SILENT- NONCODI NG	homeobox	Human Gene SWISSPROT- ID:P50219 HOMEBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	1.20E-224	7
266	cg43980506	939	TCCAAAGAAAGGGT CATGGAAGCTTA T/CjTGGGAATAAT CCTCTCAATTAGA AA	T	C				SILENT- NONCODI NG	homeobox	Human Gene TREMBLNEW- ID:G2896172 LIM HOMEBOX PROTEIN COFACTOR - HOMO SAPIENS (HUMAN), 373 aa.	1.60E-206	

267	cg43961305	100	GGGGGGTTTTTT TTTTTCTCTG[ TJTTTTTTTTTT TTTTTTTTTTTT	G	T			SILENT- NONCODI NG	hydrolase	Human Gene SWISSPROT- ID:P37980 INORGANIC PYROPHOSPHATASE (EC 3.6.1.1) (PYROPHOSPHATE PHOSPHO- HYDROLASE) (PPASE) - BOS TAURUS (BOVINE), 289 aa.	1.30E-156	10
268	cg43998672	503	CTGGGGTTTTC GGGAGGAACCA A[G/gap]GGCTCAC GGAGCCTCCTGT GCTGCA	G	gap			SILENT- NONCODI NG	hydroxyster oid	Human Gene SPTREMBL- ID:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	2.00E-220	16 (16q22)
269	cg43998672	505	GGGGGTTTTCGG GGAGGAACCAAG G[G/gap]CTCACGG AGCCTCCTGTGCT GCAGT	G	gap			SILENT- NONCODI NG	hydroxyster oid	Human Gene SPTREMBL- ID:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	2.00E-220	16 (16q22)
270	cg42908571	1031	GAGTTAATTATG TAAGTCATATT[g ap/TJATATTTTAA GAAGTACCACTTG AA	gap	T			SILENT- NONCODI NG	interleukin	Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL- 6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.	3.40E-108	7 (7p21)

271	cg42908571	1178	CTTACCTCAAATA AATGGCTAACTT[9 ap/TJATACATATT TTAAAGAAATATT A	gap	T				SILENT- NONCODI NG	interleukin	Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL- 6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.	3.40E-108	7 (7p21)
272	cg42164914	1617	CAGCCCCCATTTGT GGTCACAGGAAG[ T/CJAGAGGAGGC CACGTTCTTACTA GTT	T	C				SILENT- NONCODI NG	interleukin cept	Human Gene SWISSPROT- ID:P25025 HIGH AFFINITY INTERLEUKIN-8 RECEPTOR B (IL- 8R B) (CXCR-2) (GRO/MGSA RECEPTOR) (IL-8 RECEPTOR TYPE 2) - HOMO SAPIENS (HUMAN), 360 aa.	9.60E-191	2 (2q35)
273	cg43958501	1133	CCCAACCTGGGTT TGGCAGACATCA[ A/GJAATGATGGAG TACATTTTGCAGA TA	A	G				SILENT- NONCODI NG	isomerase	Human Gene SWISSPROT- ID:P46926 PUTATIVE GLUCOSAMINE-6-PHOSPHATE ISOMERASE (EC 5.3.1.10) (GLUCOSAMINE- 6-PHOSPHATE DEAMINASE) (OSCILLIN) (KIAA0060) - HOMO SAPIENS (HUMAN), 289 aa.	1.60E-156	5

274	cg43958501	805	CACCCCCAGGTT CTCCTAGTTCAGA [G]A/AAAAGCTGT GAAAGTGAAGA AGGA	G	A			SILENT- NONCODI NG	isomerase	Human Gene SWISSPROT- ID:P46926 PUTATIVE GLUCOSAMINE-6-PHOSPHATE ISOMERASE (EC 5.3.1.10) (GLUCOSAMINE- 6-PHOSPHATE DEAMINASE) (OSCILLIN) (KIAA0060) - HOMO SAPIENS (HUMAN), 289 aa.	1.60E-156	5
275	cg43090990	2710	TTTATTCTATTCCT ATCTGTGGATG[T/ G]GTAAATGGCTG GGGGGCCAGCCC TG	T	G			SILENT- NONCODI NG	kinase	Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1.-) (NPKC- THETA) - HOMO SAPIENS (HUMAN), 706 aa.	0	10
276	cg42879455	2259	AGCCTTTGTGCTC CCACTCAATACAIA /CJAAAAGGCCCTC TCTACATCTGGGA A	A	C			SILENT- NONCODI NG	kinase	Human Gene SWISSPROT- ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) (ATK) (B CELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa.	0	X (Xq21.3)

277	cg42879455	2283	AAAAAGGCCCTC TCTACATCTGGG A/GJATGCACCTCT TCTTTGATTCCCT GG	A	G				SILENT- NONCODI NG	kinase	Human Gene SWISSPROT- ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) (ATK) (B CELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa.	0	X (Xq21.3)
278	cg43971741	2151	AGCAACTTGGCTG AGCCCCACTACA C/TJACAGAGAAAT CATCAACCTGACT TA	C	T				SILENT- NONCODI NG	kinase	Human Gene SPTREMBL- ID:Q92749 TYPE I PHOSPHATIDYLINOSITOL-4- PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN) - HOMO SAPIENS (HUMAN), 540 aa.	1.40E-290	9
279	cg43971741	2200	TAAGAGTTTTCAA GATGTCAAACTTTC /AJAGGCTGATCAG CAGATGGGATGT GA	C	A				SILENT- NONCODI NG	kinase	Human Gene SPTREMBL- ID:Q92749 TYPE I PHOSPHATIDYLINOSITOL-4- PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN) - HOMO SAPIENS (HUMAN), 540 aa.	1.40E-290	9
280	cg43971741	2451	TTTTTAAAAATCCA TCCACACACATiga p/TJGGTAAATTAA GTATAAATTCITTT G	gap	T				SILENT- NONCODI NG	kinase	Human Gene SPTREMBL- ID:Q92749 TYPE I PHOSPHATIDYLINOSITOL-4- PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN) - HOMO SAPIENS (HUMAN), 540 aa.	1.40E-290	9





286	cg42714751	208	CCCTCCGGATTG GGCGCGCGTGCG G/C/M/JCCGCCGCG AGTGAGGGTTTTC GTGG	C	M				SILENT- NONCODI NG	kinaseinhibit or	Human Gene Similar to SWISSPROT-ID:P42771 CYCLIN- DEPENDENT KINASE 4 INHIBITOR A (CDK4I) (P16-INK4) (P16-INK4A) (MULTIPLE TUMOR SUPPRESSOR 1) (MTS1) - HOMO SAPIENS (HUMAN), 156 aa.	2.60E-53	9 (9p21)
287	cg43322545	2943	TCCAAGCTAAGCA CTGCCACTGGGG A/GJAAACTCCACC TTCCCACTTTCCC AC	A	G				SILENT- NONCODI NG	kinaserecep tor	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.lpcis:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	0	19 (19q13.1 )

288	cg43322545	3037	CCACCTCCATCCC AGACAGGTCCCTT C/GJCCCTTCTCTG TGCAGTAGCATCA CC	C	G				SILENT- NONCODI NG	kinaserecep tor	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.lpcis:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	0	19 (19q13.1 )
289	cg43322545	3038	CACCTCCATCCCA GACAGGTCCCTCT C/GJCCCTTCTCTGT GCAGTAGCATCAC CT	C	G				SILENT- NONCODI NG	kinaserecep tor	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.lpcis:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	0	19 (19q13.1 )
290	cg43980494	1040	GTCTGATAGAAGA GGAGCAGGAGAA A/GJCAAATCGTTA AAACCTAGCGAAT TC	A	G				SILENT- NONCODI NG	kinesin	Human Gene SPTREMBL- ID:Q14807 KID (KINESIN-LIKE DNA BINDING PROTEIN) - HOMO SAPIENS (HUMAN), 665 aa.	0	16



296	cg21413267	1440	CGAGCGGCACCC AGAGCCTGCACC C[T/G]CCCTCACC GTCCCTTCTGCGTC CCCC	T	G				SILENT- NONCODI NG	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	7.90E-79	
297	cg21413267	1860	AGGAGCCCTCTTC GGTGTCCTCCGAG T[C/G]CCACCGGTCA AGACCCGCAGCA CCA	T	C				SILENT- NONCODI NG	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	7.90E-79	
298	cg21413267	1890	CGGTCAAGACCC GCAGCACCAAG C[A/G]CCGCCCCC GCACCTGCCCT GTCGC	A	G				SILENT- NONCODI NG	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	7.90E-79	
299	cg42481172	1541	GAGCCGTGTGGC TGTGGCCTCCGG G[A/C]GGCGGTGG ACGGCGTGCGCT TCATC	A	C				SILENT- NONCODI NG	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	2.30E-71	1
300	cg39518465	89	GGTGCACGGCC GGCCCTGGGCAG G[gap/C]GTAGCCA TGGAGCTGTGGC GCCAAT	gap	C				SILENT- NONCODI NG	oncogene	Human Gene SWISSPROT- ID:P15498 VAV PROTO- ONCOGENE - HOMO SAPIENS (HUMAN), 846 aa.	0	

301	cg41972699	627	ATGGGGCCGGTG TCTCGCCAGGAG G[ap/C]GACAGACC CGGCTCCAGGGC CAGCGC	gap	C				SILENT- NONCODI NG	oncogene	Human Gene Similar to SWISSPROT-ID:Q64010 PROTO- ONCOGENE C-CRK (P38) (ADAPTER MOLECULE CRK) - MUS MUSCULUS (MOUSE), 304 aa.	2.40E-84	22 (22q11)
302	cg40333812	235	AGCATTTGAGGAA GCATAACTGACG[ C/T]GTGAAGGGG GTGTGGGGTACTT GCC	C	T				SILENT- NONCODI NG	oncogene	Human Gene Similar to SWISSPROT-ID:P31695 NEUROGENIC LOCUS NOTCH HOMOLOG PROTEIN 4 PRECURSOR (TRANSFORMING PROTEIN INT-3) - MUS MUSCULUS (MOUSE), 1964 aa.	1.40E-62	
303	cg43280482	2295	AGCATCTGCAGAC GACCCCCGCAGC[ A/C]TTTCCCTCGG ACCCCCCTCGAA GCC	A	C				SILENT- NONCODI NG	oncogene	Human Gene Similar to TREMBLNEW-ID:G2952331 ARG/ABL-INTERACTING PROTEIN ARGBP2A - HOMO SAPIENS (HUMAN), 666 aa.	3.90E-62	8
304	cg44014837	22	CACTGCTGTGCA GGGCAGGGA[A/T] GCTCCAGGCAGA CAGCCCCAGCAAA G	A	T				SILENT- NONCODI NG	oxidase	Human Gene SWISSNEW- ID:P08684 CYTOCHROME P450 3A4 (EC 1.14.14.1) (CYP11A4) (NIFEDIPINE OXIDASE) (NF-25) (P450-PCN1) - HOMO SAPIENS (HUMAN), 502 aa.   pcds:SWISSPROT-ID:P08684 CYTOCHROME P450 IIIA4 (EC 1.14.14.1) (NIFEDIPINE OXIDASE) (NF-25) (P450-PCN1) - HOMO SAPIENS (HUMAN), 502 aa.	8.00E-257	







312	cg42937321	1983	CCTCTTACCCCTCT CTCTCTGAACATTC /TJGTAAACAACAG ACTTACGTTAAAC T	C	T			SILENT- NONCODI NG	potassium_ channel	Human Gene SWISSPROT- ID:P22001 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.3 (HPCN3) (HGK5) (HUKIII) (HLK3) - HOMO SAPIENS (HUMAN), 523 aa.	5.40E-284	1 (1p21)
313	cg40991963	1357	CAAAATGTAACAG TGGCTTTTCAACJA /GJGGAGTAAAGCA AAGTCTCTAAAGC T	A	G			SILENT- NONCODI NG	potassium_ channel	Human Gene SWISSPROT- ID:P48048 ATP-SENSITIVE INWARD RECTIFIER POTASSIUM CHANNEL 1 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 1) (ATP-REGULATED POTASSIUM CHANNEL ROM-K) (KIR1.1) - HOMO SAPIENS (HUMAN), 391 aa.	1.80E-205	11 (11q24)

314	cg43951366	2332	AAAGATGTTTGAA TACTTAAACACTTG /AJTCACAAGATGG CAAAATGCTGAAA G	A				SILENT- NONCODI NG	prostaglandi n	Human Gene SWISSNEW- ID:P35354 PROSTAGLANDIN G/H SYNTHASE 2 PRECURSOR (EC 1.14.99.1) (CYCLOOXYGENASE -2) (COX-2) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE 2) (PROSTAGLANDIN H2 SYNTHASE 2) (PGH SYNTHASE 2) (PGHS-2) (PHS II) - HOMO SAPIENS (HUMAN), 604 aa.lpcsl:SP TREMBL- ID:Q16876 PROSTAGLANDIN ENDOPEROXIDE SYNTHASE-2 PRECURSOR (EC 1.14.99.1) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN SYNTHASE) (PROSTAGLANDIN G/H SYNTHASE) - HOMO SAPIENS (HUMAN), 604 aa.	0	1 (1q25.2)
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315	cg43951366	2829	TGGTGGAGCCAC TGCAGTGTATCTT T/C/AAAAATAAGAA TATTTTGTGAGA TA	T	C			SILENT- NONCODING	prostaglandin	Human Gene SWISSNEW- ID:P35354 PROSTAGLANDIN G/H SYNTHASE 2 PRECURSOR (EC 1.14.99.1) (CYCLOOXYGENASE -2) (COX-2) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE 2) (PROSTAGLANDIN H2 SYNTHASE 2) (PGH SYNTHASE 2) (PGHS-2) (PHS II) - HOMO SAPIENS (HUMAN), 604 aa.lpcis:SPTREMBL- ID:Q16876 PROSTAGLANDIN ENDOPEROXIDE SYNTHASE-2 PRECURSOR (EC 1.14.99.1) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN SYNTHASE) (PROSTAGLANDIN G/H SYNTHASE) - HOMO SAPIENS (HUMAN), 604 aa.	0	1 (1q25.2)
316	cg43306254	1431	CACTTAACTTGCA TGTCACAGCTT T/C/TGGTAACAAA TATCGCTAACCT TA	T	C			SILENT- NONCODING	prostaglandin	Human Gene SPTREMBL- ID:O00325 PROSTAGLANDIN EP3 RECEPTOR SUBTYPE ISOFORM - HOMO SAPIENS (HUMAN), 402 aa.	1.40E-211	1 (1p31.2)

317	cg43306254	1666	ATGTGATTAAATTA TGTGATGAAAAC[ A/T]TTTTTATAAA TGATCTTGGTCTA T	A	T			SILENT- NONCODI NG	prostaglandi n	Human Gene SP TREMBL- ID: O00325 PROSTAGLANDIN EP3 RECEPTOR SUBTYPE ISOFORM - HOMO SAPIENS (HUMAN), 402 aa.	1.40E-211	1 (1p31.2)
318	cg42918089	1064	CAATCAGAAATTGA TAAGCACTGTTCT[ C/T]TCCACTCCAT TTAGCAATTATGT CA	C	T			SILENT- NONCODI NG	protease	Human Gene Homologous to SWISSNEW-ID: P09237 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE- 7) (MMP-7) (MATRIN) - HOMO SAPIENS (HUMAN), 267 aa.   pcis: SWISSPROT-ID: P09237 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE- 7) (MMP-7) (MATRIN) - HOMO SAPIENS (HUMAN), 267 aa.	2.40E-146	11 (11q21)
319	cg44032168	1703	TCCATCCCCTCTTT TGGGCTCTTCTG[ G/C]JAGGGAAGTAA CATTACTGAGCA CC	G	C			SILENT- NONCODI NG	protease	Human Gene Similar to SWISSPROT-ID: P25155 COAGULATION FACTOR X PRECURSOR (EC 3.4.21.6) (STUART FACTOR) (VIRUS ACTIVATING PROTEASE) (VAP) - GALLUS GALLUS (CHICKEN), 475 aa.	2.40E-82	2 (2q13)

320	cg43154190	1250	TACCCGGAAGTTG AGCTCAATTTCAJT /CJTCTGTTTTCT GGCCACAACACTGC CA	T	C			SILENT- NONCODI NG	protease	Human Gene Similar to SWISSPROT-ID:P50280 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE- 7) (MMP-7) (MATRIN) - RATTUS NORVEGICUS (RAT), 267 aa.	2.40E-59	11 (11q22)
321	cg43927549	175	CCCAGTCCTGCG GCTCCTACTGGG G[A/C]GTGCGCTG GTCGGAAGATTG CTGGA	A	C			SILENT- NONCODI NG	reductase	Human Gene Homologous to SWISSPROT-ID:P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT-DIAPHORASE) (AZOREDUCTASE) (PHYLLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.	1.60E-124	6 (6pter)
322	cg43927549	191	TACTGGGGAGTG CGCTGGTCGGAA G[ap/G]ATTGCTG GACTCGCTGAAG AGAGAC	gap	G			SILENT- NONCODI NG	reductase	Human Gene Homologous to SWISSPROT-ID:P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT-DIAPHORASE) (AZOREDUCTASE) (PHYLLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.	1.60E-124	6 (6pter)

323	cg43927549	52	CGGTCCGTGGTC CCCGGGGCGCA G gap/G TCGACG GCTCCCGCCCTC CAGGCG	gap	G				SILENT- NONCODI NG	reductase	Human Gene Homologous to SWISSPROT-ID:P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT-DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.	1.60E-124	6 (6pter)
324	cg43947066	780	TTCTCAAAAGGCT GGGGGTATTTAT A/G TAAGAACTTA TTCCAAAGTGACT CT	A	G				SILENT- NONCODI NG	struct	Human Gene SWISSPROT- ID:O15142 ACTIN-LIKE PROTEIN 2 - HOMO SAPIENS (HUMAN), 394 aa.	3.30E-207	2
325	cg43923264	113	AGGAAAAGCCGGA GAATTGGGGCAC G C gap AAGAGGG GGGGCTTTGATG ACCCGC	C	gap				SILENT- NONCODI NG	struct	Human Gene SWISSPROT- ID:Q14012 CALCIUM/CALMODULIN- DEPENDENT PROTEIN KINASE TYPE I (EC 2.7.1.123) (CAM KINASE I) - HOMO SAPIENS (HUMAN), 370 aa.	1.70E-200	3
326	cg43942332	1926	AGATTCATCAGAA TAGGATTTTGC A /C AAATCCACCC ATATGCTGTTGAG C	A	C				SILENT- NONCODI NG	struct	Human Gene Homologous to SPTREMBL-ID:O00379 DELTA- CATENIN - HOMO SAPIENS (HUMAN), 792 aa.	2.10E-124	11

327	cg43274705	580	CCGCTGTCTCTGT CTTCGCITTTTAIG TTTCAAGAAGAAT AATGCGACGAAAA T	G	T				SILENT- NONCODI NG	struct	Human Gene Homologous to SPTREMBL-ID:Q28910 MUCIN - BOS TAURUS (BOVINE), 600 aa (fragment).	4.80E-110	12
328	cg42207316	146	CCACTTCTCTGGG ACACATTGCCTTIC TTTGTTTTCTCCA GCATGCGCTTGCT C	C	T				SILENT- NONCODI NG	struct	Human Gene Similar to SWISSNEW- ID:P12273 PROLACTIN-INDUCIBLE PROTEIN PRECURSOR (SECRETORY ACTIN-BINDING PROTEIN) (SABP) (GROSS CYSTIC DISEASE FLUID PROTEIN 15) (GCDFP-15) (GP17) - HOMO SAPIENS (HUMAN), 146 aa.lpcsl:SWISSPROT-ID:P12273 PROLACTIN-INDUCIBLE PROTEIN PRECURSOR (SECRETORY ACTIN-BINDING PROTEIN) (SABP) (GROSS CYSTIC DISEASE FLUID PROTEIN 15) (GCDFP-15) - HOMO SAPIENS (HUMAN), 146 aa.	3.50E-74	7 (7q32)
329	cg43927885	546	CATCATCATCATA GTTTACTTCAGC[A TTCTTAAATCCCC GAGGAGTCTGCC CT	A	T				SILENT- NONCODI NG	struct	Human Gene Similar to SWISSPROT-ID:P19065 SYNAPTOSOMAL VESICLE ASSOCIATED MEMBRANE PROTEIN 2 (VAMP-2) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 115 aa.	1.20E-55	17

330	cg40388639	5029	CTCTTGCCCAAGCC GGCTGCAAGTTT gap/TGTAAAGCGC GGGACAGACACT GCTGA	gap	T				SILENT- NONCODI NG	synthase	Human Gene SWISSPROT- ID:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.14.13.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS) - HOMO SAPIENS (HUMAN), 1434 aa.	0	12 (12q24.2 )
331	cg43949316	555	AGGTTACCAACA GGAATACAACACI C/TTCTCTCCCTT TTCTGCTCTAGAA GG	C	T				SILENT- NONCODI NG	synthase	Human Gene SWISSPROT- ID:P48651 PHOSPHATIDYL-SERINE SYNTHASE I (SERINE-EXCHANGE ENZYME I) (EC 2.7.8.-) (KIAA0024) - HOMO SAPIENS (HUMAN), 473 aa.	9.80E-269	8
332	cg43958714	1565	TGGGTGATGATCA CTGTGCTGCTTG T/CJGGCTCATGGC AGAGCATTCAGTG CC	T	C				SILENT- NONCODI NG	synthase	Human Gene Similar to SPTREMBL- ID:Q42761 SQUALENE SYNTHASE (EC 2.5.1.21) (FARNESYL- DIPHOSPHATE FARNESYLTRANSFERASE) (FARNESYLTRANSFERASE) (PRESQUALENE-DI- DIPHOSPHATE SYNTHASE) - GLYCYRRHIZA GLABRA, 412 aa.	9.20E-83	8





335	cg43275028	2601	AGAAATGGCACTGA ATTTCGTTTCITC[A] GJAACACAGATAT AATTGTTGGTTCA A	G			SILENT- NONCODI NG	synthase	Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa.jpcls:SPTREMBL-ID:P70490 O- ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	3.20E-65	1 (1q23)
336	cg43275028	2873	CTTTCACITGGTG CTGGAGAATTCA] A/GJAAAGTCAAGAA CATGCTAAGCATA AG	G			SILENT- NONCODI NG	synthase	Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa.jpcls:SPTREMBL-ID:P70490 O- ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	3.20E-65	1 (1q23)





343	cg43306266	1641	GGGATTTTGAATA TGCAATTTAAGAA[ ap/C]GTTGGGAAG AATTTACACAGATG ATG	gap	C			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.	4.80E-212	1 (1p31.2)
344	cg43306266	1650	GAATATGCATTTA AGAAAGTTGGGAA[ G/C]AATTCACAG ATGATGATTGGAG GA	G	C			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.	4.80E-212	1 (1p31.2)

345	cg43329467	683	TCGGCAAATCTTG AAAGCTGCAGGGI C/TTCAGAGACAT GGATGTGACTTCC CA	C	T			SILENT- NONCODI NG	tm7	Human Gene SWISSNEW- ID:Q99527 CHEMOKINE RECEPTOR-LIKE 2 (IL8-RELATED RECEPTOR DRY12) (FLOW- INDUCED ENDOTHELIAL G PROTEIN-COUPLED RECEPTOR) (FEG-1) (G PROTEIN-COUPLED RECEPTOR GPR30) (GPCR-BR) - HOMO SAPIENS (HUMAN), 375 aa.   pcls:SWISSPROT-ID:Q99527 CHEMOKINE RECEPTOR-LIKE 2 (IL8-RELATED RECEPTOR DRY12) (FLOW-INDUCED ENDOTHELIAL G PROTEIN-COUPLED RECEPTOR) (FEG-1) (G PROTEIN-COUPLED RECEPTOR GPR30) - HOMO SAPIENS (HUMAN), 375 aa.   pcls:TREMBLNEW-ID:G2656121 G-PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 375 aa.	8.20E-201	7
346	cg2751286	439	AAGGCATAAGAAC TAGGAGCTGCTGI gap/GJACATTTCAC TATGAAGGGCAAC TCC	gap	G			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P50052 TYPE-2 ANGIOTENSIN II RECEPTOR (AT2) - HOMO SAPIENS (HUMAN), 363 aa.	2.00E-197	X

347	cg11751407	76	GAATGTGGGGAT AAGGCATTGGGA C/C/TCTATCAGGT ATCCTGAGGAGA GACT	C	T			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P46089 PROBABLE G PROTEIN- COUPLED RECEPTOR GPR3 (ACCA ORPHAN RECEPTOR) - HOMO SAPIENS (HUMAN), 330 aa.	3.20E-176	1
348	cg43326635	135	CAGCCGGGAGCT CTGCCAGCTTTGG [C/T]GAAGGAGGG TGCTTGCCTCGTG CCC	C	T			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	1.10E-173	1
349	cg43326635	139	CGGAGCTCTGC CAGCTTTGGCGAA [G/C]GAGGGTGCT TGCCTCGTGCCC CTTG	G	C			SILENT- NONCODI NG	tm7	Human Gene SWISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	1.10E-173	1
350	cg43993798	1839	TGCTCTTGCTGCT GATGGAGGAGGA[ A/G]GGGGTGAT CCCGTGGAGCCT CCAA	A	G			SILENT- NONCODI NG	tm7	Human Gene Homologous to SWISSPROT-ID:P31421 METABOTROPIC GLUTAMATE RECEPTOR 2 PRECURSOR - RATTUS NORVEGICUS (RAT), 872 aa.	6.90E-109	3 (3q21)

351	cg43040271	2130	ATGCTTCCCCAA CCCTAGGGAATC[ A/C]ACACTTAAGA TAATTGCGCACTT CT	A	C				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa. pcls:SP TREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.	2.90E-74	
352	cg43040271	2139	CCAACCCCTAGGG AATCAACACTTAA[ G/T]ATAATTGCGC ACTTCTCCTCTTT CT	G	T				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa. pcls:SP TREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.	2.90E-74	



353	cg43040271	2163	AGATAATTGCGCA CTTCTCCTCTTTC /TTCTCTGCTCCG CTCACGGCTTGCA G	C	T				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.lpcis:SPTREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.	2.90E-74	
354	cg43040273	1668	CGCAGAGCCCCG CCGTGGGTCCGC C[T/C]GCTGAGGC GCCCCAGCCAG TGCGC	T	C				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
355	cg43040273	1760	CAGCGCCTTCTTG CTGGCACCCCAAT A/G]GAAGCCATGC GCCGGACCACGA CGT	A	G				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
356	cg43040273	1793	TGCGCCGGACCA CGACGTCACGCA G[C/G]AAAGGGAC GAGGTGTGGGTG GTGGG	C	G				SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)

357	cg43040273	2767	GCAGGTCTCTTT GAAGGCCTATGG G/CJAATGGCTACT CCAGCAACGGCA ACA	G	C			SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
358	cg43040273	2953	ATTGTAGTACAAA TGACTCACTGCTT G/AJTAAAGCAGTT TTTCTACTTTTAA G	G	A			SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
359	cg43040273	3053	ATAAACTTAGAAT AAAATTGTAAAAG ap/AJTGTATAGAG ATATGCAGAAAGGA AG	gap	A			SILENT- NONCODI NG	tm7	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	2.00E-58	5 (5q32)
360	cg43998970	1501	AGGGGTGGAAC GCTGATGGGATTT [gap/TJCCTTCATT CCTTCTGATAAAG GTA	gap	T			SILENT- NONCODI NG	transcriptfac tor	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	1.70E-177	12
361	cg43998970	249	AGCCTCCCCAGA GACAACACCGGG A/G/CJCCTCATCTC TCTCCTCACCCCTG CTG	G	C			SILENT- NONCODI NG	transcriptfac tor	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	1.70E-177	12

362	cg43947199	2623	GTC TTC TCC GCG C CCACCCGCTGGI C/TAAAGGGAAGT GGCGAAGCTGG AGC	C	T				SILENT- NONCODI NG	transcriptfac tor	Human Gene SWISSNEW- ID:P23193 TRANSCRIPTION ELONGATION FACTOR S-II (TRANSCRIPTION ELONGATION FACTOR A) - HOMO SAPIENS (HUMAN), 301 aa.[pcls:SWISSPROT-ID:P23193 TRANSCRIPTION ELONGATION FACTOR S-II (TRANSCRIPTION ELONGATION FACTOR A) - HOMO SAPIENS (HUMAN), 301 aa.	4.20E-158	8
363	cg43917396	934	GGGGCCGGGCAC TGCCAGGAAGG G[A/G]CTCCGGGA GAGGAGCCCGG GGCTG	A	G				SILENT- NONCODI NG	transcriptfac tor	Human Gene Similar to TREMBLNEW-ID:G2920821 TRANSCRIPTION FACTOR T-BOX 5 - HOMO SAPIENS (HUMAN), 518 aa.	6.90E-68	
364	cg40351913	2030	AGACGAAGACCC CAGGAAGTCATCC [T/C]GCAATGGGA GAGACACGAACA AACC	T	C				SILENT- NONCODI NG	transport	Human Gene SWISSPROT- ID:Q01959 SODIUM-DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	0	5 (5p15.3)
365	cg43921289	237	CCACGCCTGCC AGGAGCAAGCCG A[ap/A]GAGCCAG CCGCCGGCGCA CTCCGA	gap	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P02545 LAMIN A (70 KD LAMIN) - Homo sapiens (Human), 664 aa.	0	1

366	cg43928515	3196	AAACAAATAAGCC CTTTTACTGACJA /GJATGCACCCAAC CTTTTCAGCTGAA G	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:Q14687 HYPOTHETICAL PROTEIN KIAA0182 - Homo sapiens (Human), 1157 aa (fragment).	0	16
367	cg43955093	1309	AGAGTCAAAAATC CAAGTTTGGATT[C /GJTAAGCAGCCTT GACAGTAATCACT G	C	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	0	16
368	cg43955093	1336	AAGCAGCCTTGAC AGTAATCACTGAJA /GJTGGTAGGGAAA AAAAGACAGTTGG G	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	0	16
369	cg43925474	2206	AGGCCAAAAGCTCA CAGTAAATGTATJA /CJCCAGAACACAG GGCCTAAGTGAA GGT	A	C				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P42566 EPIDERMAL GROWTH FACTOR RECEPTOR SUBSTRATE SUBSTRATE 15 (PROTEIN EPS15) (AF-1P PROTEIN) - Homo sapiens (Human), 896 aa.	0	1 (1p32)
370	cg44014437	4893	CTGCTCCCANCTT CGCCAGCCTCCA[ A/GJGTACAACTT CCGCGTGTAGTG GGC	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P53675 CLATHRIN HEAVY CHAIN 2 (CLH-22) - Homo sapiens (Human), 1640 aa.	0	17 (17q11)

371	cg44014448	5114	CTGCTCCCAACTT CGCCAGCCTCCA A/GTGTACAACTT CCGCGGTAGTG GGC	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P53675 CLATHRIN HEAVY CHAIN 2 (CLH-22) - Homo sapiens (Human), 1640 aa.	0	17 (17q11)
372	cg43973129	2242	CAC TTCAC TGA A GAC ACCAT TTA TC /A/TAC CCAAGGC AGAAAGTAGAACT T	C	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P05060 SECRETORIN I PRECURSOR (SGI) (CHROMOGRANIN B) - Homo sapiens (Human), 677 aa.	0	20 (20pter)
373	cg43950657	1939	GATAGGACTCAAG CTTATTTGGGATC /T/CTGATCAATTC TTTCTGATGTTGT T	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:Q13009 T-LYMPHOMA INVASION AND METASTASIS INDUCING PROTEIN 1 (TIAM1 PROTEIN) - Homo sapiens (Human), 1591 aa.	0	21 (21q22.1 )
374	cg43956384	2416	TACAGCCATCTGT ACCTACTGGAGCI C/TJGCAGAAGGG AAGTCCACTCAGT CAC	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P13866 SODIUM/GLUCOSE COTRANSPORTER 1 (NA(+)/GLUCOSE COTRANSPORTER 1) (HIGH AFFINITY SODIUM-GLUCOSE COTRANSPORTER) - Homo sapiens (Human), 664 aa.	0	22 (22q13.1 )

375	cg43992229	101	AGCAGTGCAGCC CCGGCGCGGAGC A[G]AGGAGCCTC GGCCCGCGCCCG GCGCC	G	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P23352 KALLMANN SYNDROME PROTEIN PRECURSOR (ADHESION MOLECULE-LIKE X-LINKED) - Homo sapiens (Human), 680 aa.	0	X (Xp22.3)
376	cg44932392	260	GAGAAAAAGCATG GTACCCAAACCGA A/TTTTCCACTTTT CAGCAATACTTCA C	A	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene TREMBLNEW- ACC:AAD23581 CULLIN 2 - HOMO SAPIENS (HUMAN), 745 aa.	0	
377	cg44932392	323	TAAAGTTTAAAGA AATGTCATAATG[A /T]CATGAGCTTGA AATATCTCTAGGC A	A	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene TREMBLNEW- ACC:AAD23581 CULLIN 2 - HOMO SAPIENS (HUMAN), 745 aa.	0	
378	cg43981656	1121	AGCAAAAGAAACAC TGGCAGAAATTC[C A/T]GCATTTGCAA AATTCTAAGTTTT GG	A	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene TREMBLNEW- ACC:CAA08974 GUANINE NUCLEOTIDE-EXCHANGE FACTOR - HOMO SAPIENS (HUMAN), 548 aa.	1.60E-292	10
379	cg44910613	366	AAATAAATGTTTT CATAGTCATTAC[T /A]CTTTACAATGG GAGTGCTAAAATT C	T	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P38567 HYALURONIDASE PRECURSOR (EC 3.2.1.35) (SPERM SURFACE PROTEIN PH- 20) (SPERM ADHESION MOLECULE 1) - Homo sapiens (Human), 509 aa.	1.20E-280	7

380	cg44035104	189	AAC TGGGTTGCTC T TAAGAACTGATG T /CJCTAAACCGTCT CAGCATGGCCTG TA	C			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P37287 N- ACETYLGLUCOSAMINYL- PHOSPHATIDYLINOSITOL BIOSYNTHETIC PROTEIN (GLCNAC-PI SYNTHESIS PROTEIN) (PHOSPHATIDYLINOSITOL GLYCAN COMPLEMENTATION CLASS A) (PIG-A) - Homo sapiens (Human), 484 aa.	4.70E-261	X (Xp22.1)
381	cg43929959	1643	CAATGCATGAATC TGTAACCCCTTCGGI G/gapJAGGGCACT CACATGCCGCCCC CCAGC	gap			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SPTREMBL- ACC:P78506 DIABETES MELLITUS TYPE I AUTOANTIGEN (ISLET CELL AUTOANTIGEN P69) - HOMO SAPIENS (HUMAN), 483 aa.	2.10E-258	7
382	cg43950250	1961	TTGTTTCATGATT C TTGATGTTCTCTC /gapJTAATGAAAA CTAAGAGATGGAA TT	gap			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P11926 ORNITHINE DECARBOXYLASE (EC 4.1.1.17) (ODC) - Homo sapiens (Human), 461 aa.	7.00E-251	2 (2p25)
383	cg43064090	129	GCCGAGTCCGCT GGTGGCGGACCC CJA/TJAGGGGAGC AGCCAGTAGGGA AGTTG	T			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	4.80E-213	

384	cg43064090	130	CCGAGTCCGCTG GTGGCGGACCC A[AT]GGGAGCA GCCAGTAGGAA GTTGG	A	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	4.80E-213	
385	cg43064090	157	GGGAGCAGCCAG TAGGGAAGTTGG G[C/G]GAGTTCCA GAATCAGGGGGC GTGGC	C	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	4.80E-213	
386	cg43064090	61	TAATCGGGAGGG CTGGAGCAGAGG G[C/G]GGCCCCGC CGAGGGGCGTGG TCAGT	C	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	4.80E-213	
387	cg30490224	3296	GATGCCAAAAAA CAAAGGTGAGAA A/C]CCACAACACA GGTCTAAACTCAG CA	A	C				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P30968 GONADOTROPIN- RELEASING HORMONE RECEPTOR (GNRH-R) - Homo sapiens (Human), 328 aa.	1.20E-177	4 (4q21.2)
388	cg43924431	381	GTCTTTTACAGAT GGTTTTTCAAAA[T /gap]AGAGTCCAG TAAAAATATTTTAC ATT	T	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q16637 SURVIVAL MOTOR NEURON PROTEIN 1 - Homo sapiens (Human), 294 aa.	4.20E-166	5



389	cg43936047	607	CGTTGTTCTTAAT GTGGATCTACCAI C/TJCCCTGTGTTT ATCGAGATCCGG TC	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene TREMBLNEW- ACC:AAD40550 P38IP - HOMO SAPIENS (HUMAN), 733 aa.	4.30E-164	13
390	cg43272443	1542	TGGGATTACAGGT GCGCACTACCAI A/GJCCAAGCTAAT TTTTGTATTTT G	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SWISSNEW- ACC:P13726 TISSUE FACTOR PRECURSOR (TF) (COAGULATION FACTOR III) (THROMBOPLASTIN) (CD142 ANTIGEN) - Homo sapiens (Human), 295 aa.	7.70E-158	1 (1p22)
391	cg43966848	2065	CCTTCAGCACCCC TGCAGCGGAAA C/TJAATGAGCCGC CGTAGCCGCCAT CCG	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q92600 PROTEIN INVOLVED IN SEXUAL DEVELOPMENT, COMPLETE CDS - HOMO SAPIENS (HUMAN), 299 aa.	4.90E-156	2
392	cg43964140	176	AAAAAGCTACAGA AAAGAAATCACTT /CJTGAACAAACACA ATGACTCAGAGG CA	T	C				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to TREMBLNEW-ACC:AAC69899 SACM21 - MUS MUSCULUS (MOUSE), 721 aa.	1.10E-150	6
393	cg43285114	418	CAGGGACATGCG GGCACCCCGTGG G[G/gap]TCTTTGG CGGCTCACAGGA CAATGG	G	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to TREMBLNEW-ACC:AAD23440 LR8 - HOMO SAPIENS (HUMAN), 270 aa.	1.90E-138	7

394	cg43948566	370	GCAGGCAGAGCA CCCTGGGACCCC A[G/gap]GGCAGAA GGACCCCTGCCCTCCAGT	G	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:P18582 CD81 ANTIGEN (26 KD CELL SURFACE PROTEIN TAPA-1) - Homo sapiens (Human), 236 aa.	3.30E-125	11
395	cg44003626	649	TAAACAGCTCAGT TCAGGGACTGGT A/GJTACAAGCTGG CCACCCATCTCAG CC	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:Q15025 MRNA (HA1652) FOR ORF. PARTIAL CDS - HOMO SAPIENS (HUMAN), 296 aa (fragment).	2.70E-123	
396	cg43917206	259	TTACAGGACATCA CCTGCCATCTTA[T /A]GGTTTAATATTT ACAAATGCCTAGT	T	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P22061 PROTEIN-L-ISOASPARTATE(D- ASPARTATE) O- METHYLTRANSFERASE (EC 2.1.1.77) (PROTEIN-BETA- ASPARTATE METHYLTRANSFERASE) (PIMT) (PROTEIN L- ISOASPARTYL/D- ASPARTYL METHYLTRANSFERASE) (L- ISOASPARTYL PROTEIN CARBOXYL METHYLTRANSFERASE) - Homo sapiens (Human), 226 aa.	6.90E-118	6

397	cg43289666	215	GGCCGATTTTCC ACAAATTTAAATC[C /TJCAGTTCACCTG GTATCCAGCTCCA G	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:000559 CANCER ASSOCIATED SURFACE ANTIGEN - HOMO SAPIENS (HUMAN), 213 aa.	2.50E-111	8
398	cg43986282	840	GTTTCCACCTCCC CAGACAGGCATTI C/TJCGAGTGGGA GGCGGAGCACG TACC	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:P97314 DOUBLE LIM PROTEIN-1 - MUS MUSCULUS (MOUSE), 193 aa.	2.90E-110	12
399	cg43986282	841	TTTCCACCTCCC AGACAGGCATT[C C/TJGAGTGGGAG GCGGGAGCACGT ACCG	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:P97314 DOUBLE LIM PROTEIN-1 - MUS MUSCULUS (MOUSE), 193 aa.	2.90E-110	12
400	cg43297716	1030	CTAAACCCAAATG GGGCTGCTGGC[ A/TJGACCCCGAG GGTGCCTGGCCA GTCC	A	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P15018 LEUKEMIA INHIBITORY FACTOR PRECURSOR (LIF) (DIFFERENTIATION- STIMULATING FACTOR) (D FACTOR) (MELANOMA-DERIVED LPL INHIBITOR) (MLPLI) - Homo sapiens (Human), 202 aa.	1.20E-106	22 (22q12.1 )

401	cg43980312	2160	TTTTATCATTAAG TGCCAGAATGG[C/ T]CTTTAATGAAA ACAAAAACAAAG	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P34741 SYNDECAN-2 PRECURSOR (FIBROGLYCAN) (HEPARAN SULFATE PROTEOGLYCAN CORE PROTEIN) (HSPG) (SYND2) - Homo sapiens (Human), 201 aa.	7.90E-101	8 (8q22)
402	cg43939240	624	GGAGGGTTGGAG TCACTGACGAATG [C/T]GAGCCGGGC CAGGCCCATGCA AAGG	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O43399 HD54+INS2 ISOFORM - HOMO SAPIENS (HUMAN), 206 aa.	1.00E-100	
403	cg43941552	881	GCCACCTGCCCG GGCTGTGGAGGA G[C/gap]GCTCGCG CTGACCAGGCGC TGGGGC	C	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P11686 PULMONARY SURFACTANT-ASSOCIATED PROTEIN C PRECURSOR (SP-C) (SP5) (PULMONARY SURFACTANT-ASSOCIATED PROTEOLIPID SPL(VAL)) - Homo sapiens (Human), 197 aa.	1.60E-100	
404	cg43941552	1124	GCTTCTGCCCCACA CCGCAGGGACAA[ A/G]CCCTGGAGAA ATGGGAGCNTGG GGA	A	G				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P11686 PULMONARY SURFACTANT-ASSOCIATED PROTEIN C PRECURSOR (SP-C) (SP5) (PULMONARY SURFACTANT-ASSOCIATED PROTEOLIPID SPL(VAL)) - Homo sapiens (Human), 197 aa.	1.60E-100	

405	cg42917153	914	CATTCTCTTTGT ACATAATACATTTC /TACCTCCCTGCC TCCTCTCCTTTCT A	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P45973 HETEROCHROMATIN PROTEIN 1 HOMOLOG ALPHA (HP1 ALPHA) (ANTIGEN P25) - Homo sapiens (Human), 191 aa.	2.10E-100	12
406	cg43927693	878	CAGGGGTCAGCA GAGCTTCAGAGG TGTGTCCTCCACC TGAGCCCCCACC CGGGA	G	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P30536 PERIPHERAL-TYPE BENZODIAZEPINE RECEPTOR (PBR) (PKBS) (MITOCHONDRIAL BENZODIAZEPINE RECEPTOR) - Homo sapiens (Human), 169 aa.	5.30E-95	22
407	cg43951338	507	CAGAAAGCAGCA AATTAGTGTTTTT C/AJAGGACCGAAT TCGGCTCCCGCA GCT	C	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.	3.40E-93	10
408	cg43951338	511	AAGCAGCAAATTA GTGTTTTTCAGGJA /CJCCGAATTCGGC TCCCGCAGCTCCT G	A	C				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.	3.40E-93	10
409	cg43951338	547	CTCCCGCAGCTC CTGCATCTCCATT C/TJGTCTAGATT TATTTCTCTTTGC A	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.	3.40E-93	10

410	cg25236776	1234	CCCGCCAGCCC GAGCCTACTGA G[ap]/TCCCCGG CTCGCCCCACCG GCGCGC	gap	T			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	
411	cg25236776	1240	CCAGCCCCGACGC CTACTGAGCCCC G[C]/TGTCTCGCCC CACCGGCGGCT CTTCG	C	T			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	
412	cg25236776	1242	AGCCCCGACGCCT ACTGAGCCCCCGC G[C]/TTGCCCCCA CCGGCGCGCTCT TCGCG	C	T			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	
413	cg25236776	1246	CGACGCCCTACTG AGCCCCGCGCTC G[C]/TJCCACCGG CGCGCTCTTCGC GCCCG	C	T			SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	

414	cg43968406	1362	GCTACGTTTACTC ACAGCCAGCGAAI gap/AJCTGACATTA AAATAACTAACAA ACA	gap	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:E47283 DNA FOR ORF1 AND ORF2 FROM CHROMOSOME X - HOMO SAPIENS (HUMAN), 157 aa.	5.00E-83	X (Xp11.4)
415	cg42748886	104	CGCCTCTGATCCA AGCCACCTCCCGI C/TJCAGAGAGGTG TCATGGGCTTCCA AA	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01258 CALCITONIN PRECURSOR - Homo sapiens (Human), 141 aa.	2.00E-70	11 (11p15.2)
416	cg43969533	356	CTCTGCACAAAG GAAGCCATATCCTA [T/gap]TTTTTTTT CCTTTGCGAAAC AGA	T	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD39844 HSPC028 - HOMO SAPIENS (HUMAN), 419 aa.	1.60E-67	7
417	cg43976681	1119	AATGCCTCAGATC AGTGACCCCAAGG A/gapJACCTTCCAG AATGGATGAAATA GAC	A	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD29427 MYOMEGALIN - RATTUS NORVEGICUS (RAT), 2324 aa.	4.30E-66	11
418	cg43976681	1120	ATGCCTCAGATCA GTGACCCCAAGG A/gapJCCCTTCCAGA ATGGATGAAATAG ACC	A	gap				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD29427 MYOMEGALIN - RATTUS NORVEGICUS (RAT), 2324 aa.	4.30E-66	11

419	cg43984044	714	CCAAGCGGAAGG C CCATTTTCCTGC[ C/T]CTTCCTCAGT TGTCGGGGGCGG GGG	C	T				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O00455 TTF-1 INTERACTING PEPTIDE 20 - HOMO SAPIENS (HUMAN), 385 aa (fragment).	7.30E-66	19
420	cg43933283	398	CTAATTGTGTCGA G ATTCCAGGATT[G /A]GAGGAAAGTT GCTCCCTTTCAGC C	G	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P05062 FRUCTOSE-BISPHOSPHATE ALDOLASE B (EC 4.1.2.13) (LIVER- TYPE ALDOLASE) - Homo sapiens (Human), 363 aa.	6.60E-65	9 (9q22)
421	cg42381630	577	AAAGCAATCACAG G TGTTAAAAGAAG[ G/A]CACGTTGAAA TGATGCAGGCTG CTC	G	A				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O76087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	5.90E-64	
422	cg41664708	423	CCAGCCAGCTCAT G TTCACCTTACAC[G /C]CTCATGGACTG AGTTTACTACAC C	G	C				SILENT- NONCODI NG	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P47992 LYMPHOTACTIN PRECURSOR (CYTOKINE SCM-1) (ATAC) (LYMPHOTAXIN) (SCM-1- ALPHA) - Homo sapiens (Human), 114 aa.	2.00E-54	1



423	cg43277632	3906	AGCCTTCCCGCA GAAAAGATGCAG [C/T]CCCCCAGAC CTTCTCTGTGCTG ATT	C	T	Ala	Val (652)	CONSER VATIVE	ATPase_ associated	Human Gene SWISSPROT- ID:P35670 COPPER- TRANSPORTING ATPASE 2 (EC 3.6.1.36) (COPPER PUMP 2) (WILSON DISEASE-ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1465 aa.	0	13 (13q14.3 )
424	cg40310734	1138	TACCAGAGGCTG CATCGGCTGCGC GIC/GJAGAGCAGA TGGCGTCGTATTT TGGG	C	G	Ala	Gly (653)	CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
425	cg40310734	1238	TGGTGGGCGCTC CACTGTATATGGA [G/C]AGCCCGGCA GACCGAAAACCTG GCCG	G	C	Glu	Asp (654)	CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
426	cg40310734	1893	CTCTCAACAGGCA GGCACCCACCCTG[ A/G]ACCTGGATCT GGCGGAAAGCA CAG	A	G	Asn	Asp (655)	CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)

427	cg43982507	1883	GGTACAAGTGTG AATGTAGTCGTG G/CJCTATCAAATG GATCTTGCTACTG GC	G	C	Gly	Ala (656)	CONSER VATIVE	eph	Human Gene SWISSPROT- ID:P98155 VERY LOW-DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	0	9 (9p24)
428	cg41554010	949	GCCGAGGACGTG CGTGGCAACCTG A/G/AJGGGCAACA CCGAGGGGCTGC AGAAG	G	A	Arg	Lys (657)	CONSER VATIVE	eph	Human Gene SWISSNEW- ID:P06727 APOLOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa. lpcis:SWISSPROT-ID:P06727 APOLOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	1.80E-203	11 (11q23)
429	cg43299024	1036	TACGAGGTGCCC TTGGAGACCCCG C/A/GJTGTCACAC GCCGGGCACCGT CCCCA	A	G	His	Arg (658)	CONSER VATIVE	glucoamylase	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	7.40E-199	17 (17q25.2)
430	cg43299024	1108	GAGGAGCCCTTC GGGTGATCGTG C/A/GJCCGGCAGC TGACGGCCCGC TGCTG	A	G	His	Arg (659)	CONSER VATIVE	glucoamylase	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	7.40E-199	17 (17q25.2)

431	cg43285373	12840	GACTGACTGGGG AAAGGAACCTAAA [A/C]TCGAGTCTG CCTGGATGAATG GAGA	A	C	Ile	Leu (660)	CONSER VATIVE	glycoprotein	Human Gene SWISSPROT- ID:P98164 LOW-DENSITY LIPOPROTEIN RECEPTOR- RELATED PROTEIN 2 (MEGALIN) (GLYCOPROTEIN 330) - HOMO SAPIENS (HUMAN), 1751 aa (fragment).	0	2
432	cg36834323	1004	AGTTATTCTAGAG GATACAGAAATC] A/G]TCGAAAGTTCC CGAGAAACTAGG GAG	A	G	His	Arg (661)	CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	6.40E-91	
433	cg41568631	2101	GGACCAGGGGGC CATGCTGCTCAAT [G/A]TCTCAGGCC ACGTC AAGGAGA GCGG	G	A	Val	Ile (662)	CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	9.90E-70	14 (14q11.2 )
434	cg42359655	666	TGCTTTTCAGGGC GGAAAACCTCTCT] A/G]TTGTCCTGCG AGCTGAAGATATC CC	A	G	Ile	Val (663)	CONSER VATIVE	hydrolase	Human Gene SWISSPROT- ID:P09848 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYL CERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa.	0	2 (2q21)



437	cg43933479	133	AAGGAGAAAGAGA AAGCTGTTTATCC[ A/GJTTCCATGGGT GAAGGTACAATAA AT	A	G	His	Arg (666)	CONSER VATIVE	interleukin	Human Gene SWISSNEW- ID:P29466 INTERLEUKIN-1 BETA CONVERTASE PRECURSOR (IL- 1BC) (EC 3.4.22.36) (IL-1 BETA CONVERTING ENZYME) (ICE) (INTERLEUKIN-1 BETA CONVERTING ENZYME) (P45) (CASPASE-1) (CASP-1) - HOMO SAPIENS (HUMAN), 404 aa.lpcis:SWISSPROT-ID:P29466 INTERLEUKIN-1 BETA CONVERTASE PRECURSOR (IL- 1BC) (EC 3.4.22.36) (IL-1 BETA CONVERTING ENZYME) (ICE) (INTERLEUKIN-1 BETA CONVERTING ENZYME) (P45) (CASPASE-1) (CASP-1) - HOMO SAPIENS (HUMAN), 404 aa.	2.50E-206	
438	cg43942537	1163	GCCACTGTCTCTT CCAAACCCCTTCA[ C/AJGCCCTTGCTT GCTTGTCTCGTC TA	C	A	Val	Leu (667)	CONSER VATIVE	kinesin	Human Gene SWISSNEW- ID:P33176 KINESIN HEAVY CHAIN (UBIQUITOUS KINESIN HEAVY CHAIN) (UKHC) - HOMO SAPIENS (HUMAN), 963 aa.lpcis:SWISSPROT-ID:P33176 KINESIN HEAVY CHAIN (UBIQUITOUS KINESIN HEAVY CHAIN) (UKHC) - HOMO SAPIENS (HUMAN), 963 aa.	0	10

439	cg38337333	1035	TTCCAAATGCTGA GCCCAGAGCGTTI G/ATCTCCTGCCC ATGAGCACCCACA GTC	G	A	Val	Ile (668)	CONSER VATIVE	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
440	cg38337333	271	CTGGAACAGTTTC CTCATTAGCCCTI G/CTGACCCAG CACACGCAGGGA CCTA	G	C	Val	Leu (669)	CONSER VATIVE	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
441	cg38337333	823	TCATCGCTGGTGC TCCAAAAAATA /GJATGCTGCTGTA ATGAACCAAGAGC C	A	G	Asn	Asp (670)	CONSER VATIVE	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
442	cg30421838	3434	GGATGCTGTTGCT CTCCACAGCCAI G/TTGGCGTTCC AAATGAAAGCCAA GC	G	T	Val	Leu (671)	CONSER VATIVE	nucl_recpt	Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.   pcls:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.	0	11 (11q22)

443	cg43064060	1019	GCCAATGGCATC CAGAACAGGAG G/C/TJGGAGTCC GCATCTTTCAC TG CTGC	C	T	Ala	Val (672)	CONSER VATIVE	nucl_recpt	Human Gene SWISSPROT- ID:Q07869 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA (PPAR-ALPHA) - HOMO SAPIENS (HUMAN), 468 aa.[pcls:SPTREMBL-ID:Q16241 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA - HOMO SAPIENS (HUMAN), 468 aa (fragment).	4.10E-254	22
444	cg43991813	1860	TCTCGACTAACAG CATTTCCAAAGAT /CJGGAGCGAATAT TGTCACCGGTTGA G	T	C	Ile	Val (673)	CONSER VATIVE	nuclease	Human Gene SWISSPROT- ID:P40692 MUTL PROTEIN HOMOLOG 1 (DNA MISMATCH REPAIR PROTEIN MLH1) - HOMO SAPIENS (HUMAN), 756 aa.	0	3 (3p21.3)
445	cg42904626	194	GAGTGCCCTTGAC GATACAGCTAATT C/GJAGAATCATTT TGTTGGACGAATAT GA	C	G	Gln	Glu (674)	CONSER VATIVE	oncogene	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K- RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	1.10E-97	12
446	cg42904626	548	AAGAAGTTATGGA ATTCCCTTTTATT[G/ CJAAACATCAGCA AAGACAAGACAG GG	G	C	Glu	Gln (675)	CONSER VATIVE	oncogene	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K- RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	1.10E-97	12

447	cg42460457	2845	GCCGCCCTCAGCC AGCAAGCAGGCG G[C/T]TAGGCCAG TCCTAGCCACCAC AGAG	C	T	Ala	Val (676)	CONSER VATIVE	phosphate	Human Gene SWISSPROT- ID:P23470 PROTEIN-TYROSINE PHOSPHATASE GAMMA PRECURSOR (EC 3.1.3.48) (R-PTP- GAMMA) - HOMO SAPIENS (HUMAN), 1445 aa.	0	3 (3p14.2)
448	cg43272594	582	GGGATGTACTGC ATGGTGTCTTGG [T/C]GCTGTATGTG CAGGCACGACTC TGT	T	C	Val	Ala (677)	CONSER VATIVE	phosphate	Human Gene Similar to SPTREMBL- ID:Q61469 PHOSPHATIDIC ACID PHOSPHATASE - MUS MUSCULUS (MOUSE), 283 aa.	1.40E-79	19
449	cg43958858	807	TCAGGTGGTGGG AACCTACCGTTGC [C/G]TTCCTGGAA AGAAGGGAGGCT ACAC	C	G	Leu	Val (678)	CONSER VATIVE	polymerase	Human Gene SWISSNEW- ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN P1) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa.   pcds:SWISSPROT-ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN P1) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa.	0	6 (6p12)



450	cg43916732	540	GTACAGCGGGCG GGCCACCTCGGG C[AT]CTGAGCAC CAATTTGCGGGG GGCG	A	T	Thr	Ser (679)	CONSER VATIVE	protease	Human Gene SPTREMBL- ID:Q15113 PROCOLLAGEN C- PROTEINASE ENHANCER PROTEIN PRECURSOR - HOMO SAPIENS (HUMAN), 449 aa.	1.20E-247	7 (7q21.3)
451	cg42894809	2745	GGATGCTGGAGA GTGGATCACTGTC [A/G]ATCAGACGA CAACAGCCCAACC GTTA	A	G	Asn	Asp (680)	CONSER VATIVE	struct	Human Gene SWISSPROT- ID:P54296 M-PROTEIN (165 KD TITIN-ASSOCIATED PROTEIN) (165 KD CONNECTIN- ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1465 aa.	0	8
452	cg40388639	2337	GATTCCTCCAGAG CTGGTGTTGGAA[ G/C]TTCCTCATCAG GCACCCCAAGTTT GA	G	C	Val	Leu (681)	CONSER VATIVE	synthase	Human Gene SWISSPROT- ID:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.14.13.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS) - HOMO SAPIENS (HUMAN), 1434 aa.	0	12 (12q24.2)
453	cg40388639	2380	AAGTTTGAGTGGT TCAAGGACCTGGI G/C]GCTGAAGTG GTACGGCCCTCCC CGCC	G	C	Gly	Ala (682)	CONSER VATIVE	synthase	Human Gene SWISSPROT- ID:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.14.13.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS) - HOMO SAPIENS (HUMAN), 1434 aa.	0	12 (12q24.2)

454	cg43124627	1524	AATTTCTATATCA CTGGGGACACAGAG C/GJATATATGGAT AAAGATGGGTATT TC	C	G	Ala	Gly (683)	CONSER VATIVE	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.jpcl:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.70E-79	16
455	cg43124627	869	TGGAACAAGTGG ATATCCGAAATG A/TJCTGCACACAC CCACAGCAGTTTT GG	A	T	Thr	Ser (684)	CONSER VATIVE	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.jpcl:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.70E-79	16

456	cg43064068	1464	AGGAGAGGTGGT GAAGGCATTGTG [G/A]TCTGCGCT CGCAGTTCCTGTC CCA	A	Val	Ile (685)	CONSER VATIVE	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.pclis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.40E-65	
457	cg2514276	1090	GTGATGGACCCCT CTCATATATGCCTI A/TCCGCAGCCAA GAGATGCGGAAG ACC	T	Tyr	Phe (686)	CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P33032 MELANOCORTIN-5 RECEPTOR (MC5-R) (MC-2) - HOMO SAPIENS (HUMAN), 325 aa.	7.00E-172	
458	cg32423505	964	TTCCATCTGAGGT TTATAAACACG/A T/ATTTCAGGCAA GTGCCAGAAATG GC	T	Phe	Tyr (687)	CONSER VATIVE	tm7	Human Gene Similar to SPTREMBL- ID:Q89609 G PROTEIN-COUPLED RECEPTOR - EQUINE HERPESVIRUS TYPE 2 (EHV-2), 383 aa.	1.20E-55	3 (3q21)
459	cg43335558	344	CAAGACCTAGCTC CCCAGCAGAGAGI C/TGGCCCCACAA CAAAAGAGGTCCA GC	T	Ala	Val (688)	CONSER VATIVE	tnfreceptor	Human Gene Similar to TREMBLNEW-ID:G2653845 TNF RECEPTOR-RELATED RECEPTOR FOR TRAIL - HOMO SAPIENS (HUMAN), 386 aa.	5.50E-89	8

460	cg43998970	1347	GACAGAGCTGTA CCGTGACATTTC C/GJAGCACCTTCG GGATGAATCAGG CAA	C	G	Gln	Glu (689)	CONSER VATIVE	transcriptfac tor	Human Gene SP TREMBL- ID: Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	1.70E-177	12
461	cg2537639	800	GAGGGCGATTCT ACTACCTGGGGI G/CJGTTCTTCGGG GGTCCGGTGCAA GAG	G	C	Gly	Ala (690)	CONSER VATIVE	transferase	Human Gene SWISSPROT- ID: P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)

462	cg43935995	1552	AGTGTCCTCACC ATGGTCACCCCTG[ A/G]TCACCCCTGCC TCTGCTTTTCCTT CT	A	G	Ile	Val (691)	CONSER VATIVE	transport	Human Gene SWISSPROT- ID:Q03518 ANTIGEN PEPTIDE TRANSPORTER 1 (APT1) (PEPTIDE TRANSPORTER TAP1) (PEPTIDE TRANSPORTER PSF1) (PEPTIDE SUPPLY FACTOR 1) (PSF-1) (PEPTIDE TRANSPORTER INVOLVED IN ANTIGEN PROCESSING 1) - HOMO SAPIENS (HUMAN), 748 aa.	0	6
463	cg43935986	1424	CCTGGAACGCGC CTTGACCTGCTC [G/A]TAAGGAGGG TGCTGCACCTTG GGGT	G	A	Val	Ile (692)	CONSER VATIVE	transport	Human Gene SPTREMBL- ID:Q28437 ABC-TRANSPORTER - GORILLA GORILLA GORILLA (LOWLAND GORILLA), 703 aa.	0	6 (6p21.3)
464	cg43968274	730	GAGCACGAGGAA GCCATGAATGCG G[C/T]CTACTCAG GCTACGCTCTACAC GCAC	C	T	Ala	Val (693)	CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:O14914 NEURONAL MUNC18- 1 BINDING PROTEIN - HOMO SAPIENS (HUMAN), 837 aa.	0	9
465	cg44018598	3568	AGATACCTTTCTAT AAGCAGTTTTTTA[G /C]ATTGTAGGAAG CAGCTGAATTCAA A	G	C	Leu	Val (694)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P29374 RETINOBLASTOMA BINDING PROTEIN 1 (RBBP-1) - Homo sapiens (Human), 1257 aa.	0	14

466	cg44926796	1825	ACACTGGAAGCA CAACAGTTGGCA] C/GJTCTGCTAG AAATAATAATTG CA	C	G	Thr	Ser (695)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:Q15046 LYSYL-TRNA SYNTHETASE (EC 6.1.1.6) (LYSINE-TRNA LIGASE) (LYSRS) (KIAA0070) - Homo sapiens (Human), 597 aa.	0	16
467	cg43055918	1622	AACGCTGCCCTG ACTGAGAAAGGC A/C/TJGATGCTCG CTCCACTGCTGGA ACCG	C	T	Arg	His (696)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	0	17
468	cg43966985	1381	CATCCAGGACAAAC TTC TCGGTGACT] C/GJAAGTGCCCTT CACTGAGAGCGC CTG	C	G	Gln	Glu (697)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P01019 ANGIOTENSINOGEN PRECURSOR - Homo sapiens (Human), 485 aa.	3.90E-257	1 (1q42)
469	cg43918854	966	CTTCAACCCTGGT CGGAGACACACGG] A/CJTCACCATGGC CATCAGAACAGTG CG	A	C	Ile	Leu (698)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P20062 TRANSCOBALAMIN II PRECURSOR - Homo sapiens (Human), 427 aa.	3.30E-228	22 (22q11.2 )
470	cg43918484	1148	CTGATTCTTCCGT TCTTCTTGACTTJC /GJTGCCACCTTGC CAGCCAGCTGCT CG	C	G	Glu	Gln (699)	CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P05089 ARGINASE 1 (EC 3.5.3.1) (LIVER-TYPE ARGINASE) - Homo sapiens (Human), 322 aa.	1.30E-171	6 (6q23)

471	cg43942977	1009	ACGGCCCTGGAG AACCAGAAAGAG G[C/T]GAGGAAGA AGAAAGTCTTGAT TGCC	C	T	Ala	Val (700)	CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	9.60E-148	
472	cg43942977	725	GGATGGTGCTG ATGAGGAGTTGG A[G/T]CAGATGCT GGACAGTGGGCA AAGCG	G	T	Glu	Asp (701)	CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	9.60E-148	
473	cg43943361	921	TGGGGTTGGCTT GGTTTCAATAAG[ G/C]AACGGGGAC ACTTACAAATTGC TGC	G	C	Glu	Gln (702)	CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:P04179 SUPEROXIDE DISMUTASE [MN] PRECURSOR (EC 1.15.1.1) - Homo sapiens (Human), 222 aa.	5.70E-124	6 (6q25.3)
474	cg25236776	1094	GTGACCGAGCCC GAGTGCCGCGAG G[G/T]CTTTACAC GCCGCGCCCGCG CCAGC	G	T	Gly	Val (703)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	

475	cg25236776	881	CAGTGCCTCCCTG GCGGCCCGGG [G/T]CAAAGGCCG CTGCTTCGGCC CAGC	G	T	Gly	Val (704)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:P01185 VASOPRESSIN- NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	7.20E-91	
476	cg38899722	30	GGCCAACTCTGCT ATGGACACCAGA G/C]TACTCTGCTG TGCGGTCATCTGT CT	G	C	Val	Leu (705)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G292791 T- CELL RECEPTOR BETA PRECURSOR - HOMO SAPIENS (HUMAN), 145 aa (fragment).	5.70E-75	
477	cg11753818	253	GCCTGGAACACC AGGCTCCTCTGC C/G/A]TGTCATGCT TTGTCTCCTGGGA GCA	G	A	Arg	His (706)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G2104755 T CELL RECEPTOR V-BETA 23 - HOMO SAPIENS (HUMAN), 129 aa (fragment).	1.30E-66	7
478	cg2526759	519	AGCCACCCAGAC CGGAGACTCGGC C/G/A]TCTACCTCT GTGCTGTGGAGG CCTA	G	A	Val	Ile (707)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	
479	cg2526759	539	CGGCCGTCTACC TCTGTGCTGTGGA [G/C]GCCCTATTCTA ACGACTACAAGCT CA	G	C	Glu	Asp (708)	CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	





482	cg43252813	2306	TGTAATTCCTGTAA TGGGGCTGATGA C/TATATATGATG GTTATGGACCACC AC	C	T	Thr	Ile (711)	NON- CONSER VATIVE	ATPase_as sociated	Human Gene SWISSNEW- ID:Q04656 COPPER- TRANSPORTING ATPASE 1 (EC 3.6.1.36) (COPPER PUMP 1) (MENKES DISEASE-ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1500 aa.lpcis:SWISSPROT-ID:Q04656 COPPER-TRANSPORTING ATPASE 1 (EC 3.6.1.36) (COPPER PUMP 1) (MENKES DISEASE- ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1500 aa.	0	X (Xq12)
483	cg43920913	929	GCCCCGTGAGCAG TCAGGACCCCGC TTC/TCCGTCCTG GAGTGCCACGAT CCCAG	C	T	Pro	Ser (712)	NON- CONSER VATIVE	biotindep	Human Gene SWISSPROT- ID:P05166 PROPIONYL-COA CARBOXYLASE BETA CHAIN PRECURSOR (EC 6.4.1.3) (PCCASE) (PROPANOYL- COA:CARBON DIOXIDE LIGASE) - HOMO SAPIENS (HUMAN), 539 aa.	8.20E-288	3 (3q21)
484	cg40310734	267	GGAGTGGGTGCT GCTGCTCTTGGG A/C/G]CTTGTGCT GCCCCCTCCAGCC TGGGC	C	G	Pro	Ala (713)	NON- CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIb) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)

485	cg40310734	3111	CGTGTCCCTCCCTC CCCTATGCGGTG C/G CCCCGCTCA GCCTGCCCCCGAG GGGA	C	G	Pro	Ala (714)	NON- CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIb) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
486	cg43956560	777	GGGTACTATGG GCCCCAGTGTC G T/C TTGTGATTC AGTGTGAGCCTTT GGA	T	C	Phe	Leu (715)	NON- CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90-MEL) (LEUKOCYTE-ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAM1) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	1.00E-218	1 (1q23)

487	cg43956560	837	GCTGGGTACCAT GGACTGTACTCAG [C/T]CTTTGGGAAA CTTCAGCTTCAGC TC	C	T	Pro	Ser (716)	NON- CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90-MEL) (LEUKOCYTE-ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAM1) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	1.00E-218	1 (1q23)
488	cg42388009	753	TGCAGAAGGCAC CACAGAGACCGG A/A/GGGCAGGGC AAGGGCACCTCG AAGAC	A	G	Arg	Gly (717)	NON- CONSER VATIVE	cadherin	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL- BINDING SIALOPROTEIN) (INTEGRIN-BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	7.00E-172	4
489	cg43977436	1945	GTGTGTGTGTAAT GGTGTGGCTGTAT C/TTGCTCCAACCA AGATCTTATTACT GA	C	T	Arg	Cys (718)	NON- CONSER VATIVE	calcium_cha nnel	Human Gene SWISSPROT- ID:P21817 RYANODINE RECEPTOR, SKELETAL MUSCLE (SKELETAL MUSCLE CALCIUM RELEASE CHANNEL) - HOMO SAPIENS (HUMAN), 5032 aa.	0	

490	cg43280376	1130	CGGAAGCTGGTG TCCTACTGCCCC [A/G]AAGGTTGCA ACAACTGTTGCCC CTC	A	G	Gln	Arg (719)	NON- CONSER VATIVE	carboxylase	Human Gene SWISSPROT- ID:P38435 VITAMIN K-DEPENDENT GAMMA-CARBOXYLASE (EC 6.4.-.- ) (GAMMA-GLUTAMYL CARBOXYLASE) - HOMO SAPIENS (HUMAN), 758 aa.	0	2
491	cg42201364	1595	CCAGGGCCTCCA GGTCCAAGAGGC C[A/C]CTCTGGAG AGCCTGGTCTTCC AGGG	A	C	Trp	Gly (720)	NON- CONSER VATIVE	collagen	Human Gene SWISSPROT- ID:Q03692 COLLAGEN ALPHA 1(X) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 680 aa.	0	6
492	cg42201364	176	GTGTTTACGCTG AACGATACCAA C/T]GCCACACAG CATAAAAGGCCCA CTA	C	T	Thr	Met (721)	NON- CONSER VATIVE	collagen	Human Gene SWISSPROT- ID:Q03692 COLLAGEN ALPHA 1(X) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 680 aa.	0	6
493	cg40339378	2855	TCCAGGAATACCA GGTCTGCCTGGT A/G]TTCCTGGAAC AAGAGGATTAAA GG	A	G	Ile	Thr (722)	NON- CONSER VATIVE	collagen	Human Gene SPTREMBL- ID:Q12823 A TYPE IV COLLAGEN - HOMO SAPIENS (HUMAN), 1690 aa (fragment).	0	X (Xq22)

494	cg43063256	606	AGACTGTGTTACC AACAGACCATGC[ A/G]GAAGTCAAGT GCGATGTGAAGG CTT	A	G	Arg	Gly (723)	NON- CONSER VATIVE	complement	Human Gene SWISSNEW- ID:P07358 COMPLEMENT COMPONENT C8 BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 591 aa. pcis:SWISSPROT-ID:P07358 COMPLEMENT COMPONENT C8 BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 591 aa.	0	1 (1p32)
495	cg44032748	414	CTCCAGTTCTACA ACTTGTGTAAGG[ A/C]AAGCACAGTG TGGACAGGATTTC CA	A	C	Lys	Gln (724)	NON- CONSER VATIVE	complement	Human Gene SWISSPROT- ID:P07357 COMPLEMENT COMPONENT C8 ALPHA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 584 aa.	0	1 (1p32)
496	cg43049885	533	CAGTTTGGGGA CAGCCATGCACT G[A/C]GCCTCTGG TAGCCTTTCAACC ATGC	A	C	Glu	Ala (725)	NON- CONSER VATIVE	complement	Human Gene TREMBLNEW- ID:G386348 COMPLEMENT C6 - HOMO SAPIENS, 941 aa.	0	5 (5p13)
497	cg21644442	1347	CCAGGCTCTCCC AGGATCTCATCAC [T/C]GCGCCCCCA GGGCTCAGCAA CCCC	T	C	Leu	Pro (726)	NON- CONSER VATIVE	csf	Human Gene SWISSPROT- ID:P09603 MACROPHAGE COLONY STIMULATING FACTOR-1 PRECURSOR (CSF-1) (MCSF) - HOMO SAPIENS (HUMAN), 554 aa.	5.00E-304	1 (1p21)



500	cg44017721	174	TTGGTAGGGACG GAACTCGGGCG C[G]TGGCGGTGG CCCGAGTGGAGA TAGGA	G	T	Pro	Gln (729)	NON- CONSER VATIVE	cytochrome	Human Gene Similar to SPTREMBL- ID:O00761 CYTOCHROME OXIDASE SUBUNIT VIA HEART ISOFORM PRECURSOR (EC 1.9.3.1) (CYTOCHROME-C OXIDASE) (CYTOCHROME A(3)) (CYTOCHROME AA(3)) - HOMO SAPIENS (HUMAN), 97 aa.	2.40E-52	22
501	cg41626024	279	GCTGGTTTGCTCC CAGGAGGCCAAG[ A/C]AGTCAGCCTA CTGCCCTACAGT CA	A	C	Lys	Gln (730)	NON- CONSER VATIVE	deaminase	Human Gene Similar to SWISSPROT-ID:P32320 CYTIDINE DEAMINASE (EC 3.5.4.5) (CYTIDINE AMINOHYDROLASE) - HOMO SAPIENS (HUMAN), 146 aa.[pcis:TREMBLNEW-ID:E1228801 CYTIDINE DEAMINASE (EC 3.5.4.5) - HOMO SAPIENS (HUMAN), 146 aa.	8.80E-78	1 (1p36.2)
502	cg43057018	1618	AAGCATCCGAACA ATCCTCATCTTTT[ G]GAAGATGCCAG GAGCAATTCGGAA T	T	G	End	Gly (731)	NON- CONSER VATIVE	dehydrogenase	Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.[pcis:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.	1.30E-209	4 (4q22)



503	cg42837709	464	CCGCACCAACGC CGACATCATCGAG [A/G]CCCTGAGGA AGAAAGGCTTCAA GGG	A	G	Thr	Ala (732)	NON- CONSER VATIVE	dna_rna_bi nd	Human Gene Similar to TREMBLNEW-ID:G913312 DNA BINDING PROTEIN MEF2 {CLONE XMEF2A1} - XENOPUS LAEVIS, 516 aa.	3.90E-86	1
504	cg43327954	2205	TCCACGACCGGG TAGAGAACTACAA [C/A]CCGCGGCAG CGCAAGCTCCGC AACC	C	A	Asn	Lys (733)	NON- CONSER VATIVE	dna_rna_bi nd	Human Gene Similar to SPTREMBL- ID:Q61491 DNA-BINDING PROTEIN - MUS MUSCULUS (MOUSE), 546 aa.	5.50E-57	1
505	cg43971258	707	TCGTTGGAGATGA CAAGTTCGGAGI C/TJGAGCTCGGCT GTCTGGATGGGA AGG	C	T	Ala	Thr (734)	NON- CONSER VATIVE	dna_rna_bi nd_inhib	Human Gene Similar to SWISSNEW- ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX- LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa. pcls:SWISSPROT-ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX-LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa.	1.30E-60	1 (1p36.13 )

506	cg41554010	1253	AGCTGGAGCAAC AGCAGGAACAGC A[G/T]CAGGAGCA GCAGCAGGAGCA GGTGC	G	T	Gln	His (735)	NON- CONSER VATIVE	eph	Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.   pcls:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	1.80E-203	11 (11q23)
507	cg43957743	1063	GTTTGGCATACT GGATATTTTAATTC /TTCAGTGGAGATA AAAGACAGCCCCA CT	C	T	Gly	Glu (736)	NON- CONSER VATIVE	esterase	Human Gene SWISSNEW- ID:Q15166 SERUM PARAOXONASE/ARYLESTERASE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDIAKYLPHOSPHATASE 3) (A- ESTERASE 3) (AROMATIC ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment).   pcls:SWISSPROT- ID:Q15166 SERUM PARAOXONASE/ARYLESTERASE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDIAKYLPHOSPHATASE 3) (A- ESTERASE 3) (AROMATIC ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment).	1.90E-178	

508	cg43957743	1079	TATTTTAATCCAG TGGAGATAAAAG[ A/C]CAGCCCACTA GGAAGTATATCAA TA	A	C	Ser	Ala (737)	NON- CONSER VATIVE	esterase	Human Gene SWISSNEW- ID:Q15166 SERUM PARAOXONASE/ARYLESTERASE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM) ARYLDIAKYLPHOSPHATASE 3) (A- ESTERASE 3) (AROMATIC ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment). pcis:SWISSPROT- ID:Q15166 SERUM PARAOXONASE/ARYLESTERASE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM) ARYLDIAKYLPHOSPHATASE 3) (A- ESTERASE 3) (AROMATIC ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment).	1.90E-178	
509	cg43248101	812	AAGTGAATTCAT CTTGCAATGAAC[ A/G]AGGAAGGAAA ACTCTATGCAAAG AA	A	G	Lys	Glu (738)	NON- CONSER VATIVE	fgf	Human Gene Homologous to SWISSPROT-ID:P21781 KERATINOCYTE GROWTH FACTOR PRECURSOR (KGF) (FIBROBLAST GROWTH FACTOR- 7) (FGF-7) (HBGF-7) - HOMO SAPIENS (HUMAN), 194 aa.	9.30E-106	15 (15q15)

510	cg43969014	332	GATGAGCTCTCCA ACCACGTATTTTC /AJTGCGTTTTTGA TCCAGACCCAGAT G	A	Arg	Ile (739)	NON- CONSER VATIVE	glucuronida se	Human Gene Similar to SWISSPROT-ID:P08236 BETA- GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa.	7.40E-80	5
511	cg43286488	387	CACCAGCAAGAT GCCACGATCAG C[G/C]GAACCTGC CCAAGGCCCTGCTT CTTG	C	Pro	Arg (740)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSNEW- ID:P40967 MELANOCYTE PROTEIN PMEL 17 PRECURSOR (MELANOCYTE LINEAGE- SPECIFIC ANTIGEN GP100) (MELANOMA-ASSOCIATED ME20 ANTIGEN) (ME20M/ME20S) (ME20- M/ME20-S) (95 KD MELANOCYTE- SPECIFIC SECRETED GLYCOPROTEIN) - HOMO SAPIENS (HUMAN), 661 aa.   pcds:SWISSPROT-ID:P40967 MELANOCYTE PROTEIN PMEL 17 PRECURSOR (MELANOCYTE LINEAGE-SPECIFIC ANTIGEN GP100) (MELANOMA-ASSOCIATED ME20 ANTIGEN) (ME20M) (ME20-M / ME20-S) (95 KD MELANOCYTE- SPECIFIC SECRETED GLYCOPROTEIN) - HOMO SAPIENS (HUMAN), 661 aa.	0	12

512	cg44004239	663	TTTTCCCCAGGG TCACAGACTGAT A/GJACCCACAGAG GTCAGGGTCTTCT GT	A	G	Tyr	His (741)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	0	
513	cg44004239	672	GGGGTCACAGAC TGATAACCCACAG [A/G]GGTCAGGGT CTTCTGTCCAGTG GTC	A	G	Ser	Pro (742)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	0	
514	cg44004239	773	CTGATGACCCACA GAAGTCATGGTC A/GJTGCCCCAGT GATCTCAGTCTTC TC	A	G	Met	Thr (743)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	0	

515	cg43932434	1504	ATATGTGTCATAC TGGGAGGTGTTG[ G/T]ATGTGAGGAT GTACACCCCTGTG TT	G	T	Ser	Tyr (744)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYtic GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HEPARAN SULFATE PROTEOGLYCAN) (EPICAN) (CDW44) - HOMO SAPIENS (HUMAN), 742 aa.	1.80E-195	11 (11pter)
516	cg40915005	622	AAGGAGCCCTCTCT CCTTCCATGTCA[ C/T]CTGGATCGCA TCCTTTTACAACC AT	C	T	Thr	Ile (745)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSNEW- ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T- CELL SURFACE ANTIGEN T6/LEU- 6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa. lpcis:SWISSPROT-ID:P06126 T- CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.	2.00E-183	1 (1q21)

517	cg40915005	737	ATTCCAGCACCAT CGTTTTCCTGTGf G/CJCCCTGGTCCA GGGAAACTTCA GCA	G	C	Trp	Cys (746)	NON- CONSER VATIVE	glycoprotein	Human Gene SWISSNEW- ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T- CELL SURFACE ANTIGEN T6/LEU- 6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.lpcis:SWISSPROT-ID:P06126 T- CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.	2.00E-183	1 (1q21)
518	cg36834323	1529	GIGCTCCCTGATC CTCGTGAAGCATf A/GJTGCTAGCTCA AGTTATGTGGCAT CT	A	G	Tyr	Cys (747)	NON- CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	6.40E-91	
519	cg36834323	329	AATGCTGCGAAAG ATATGAATGGAJA /CJGCTTTTGCATG GAAAAGCAATAAA A	A	C	Lys	Thr (748)	NON- CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	6.40E-91	

520	cg36834323	463	AAGTCTGAGATCT GCAAGAGGAAGC[ A/C]GTGGAGGAA CAAGAGGGTGGC TTCC	A	C	Ser	Arg (749)	NON- CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	6.40E-91	
521	cg44019290	1697	GCGGATAAGTAG AGGACCTTCATGT [T/G]GTATTGCTG GTGAAGTTGGTTC GG	T	G	Asn	His (750)	NON- CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:P04216 THY-1 MEMBRANE GLYCOPROTEIN PRECURSOR (THY-1 ANTIGEN) (CDW90) (CD90 ANTIGEN) - HOMO SAPIENS (HUMAN), 161 aa.	2.50E-80	11
522	cg42336656	1665	CTTAGACATACAA TATACTTACCTT[A/ G]GAGGTCACGTA TGTTTGTCCGCAC A	A	G	Arg	Gly (751)	NON- CONSER VATIVE	glycoprotein	Human Gene Similar to SWISSPROT-ID:Q05910 CELL SURFACE ANTIGEN MS2 PRECURSOR (EC 3.4.24.-) (MACROPHAGE CYSTEINE- RICH GLYCOPROTEIN) (CD156 ANTIGEN) - MUS MUSCULUS (MOUSE), 826 aa.	9.40E-58	
523	cg42730678	980	GGAGCGAGCGTG GATCCAGTTCGC G[G/T]CGGGGTG TTTGGGTCAAGTT GCTG	G	T	Ala	Asp (752)	NON- CONSER VATIVE	homeobox	Human Gene SWISSPROT- ID:P28356 HOMEBOX PROTEIN HOX-D9 (HOX-4C) (HOX-5.2) - HOMO SAPIENS (HUMAN), 342 aa.	2.60E-188	2



524	cg42714160	769	GCCCTGTGCCCTG ACGGAGAGGCAG A/T/G/CAAGATATG GTTCCAGAACCAG CGC	T	G	Ile (753)	Ser (753)	NON- CONSER VATIVE	homeobox	Human Gene Homologous to SWISSPROT-ID:P17509 HOMEBOX PROTEIN HOX-B6 (HOX-2B) (HOX-2.2) (HU-2) - HOMO SAPIENS (HUMAN), 224 aa.	1.10E-123	
525	cg42359655	3297	CTGGGCACCATAT AGGATAGCCCAC[ A/G]CCGTCATCAA AGCCCATGCCAG AGT	A	G	Thr (754)	Ala (754)	NON- CONSER VATIVE	hydrolase	Human Gene SWISSPROT- ID:P09848 LACTASE-PHILORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLKERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa.	0	2 (2q21)
526	cg43925670	2172	GTGGAGGGTGCA GGTGAAGTAGCAT [C/G]CACTTCCTTC TTCCTCTTCTTG AT	C	G	Asp	His (755)	NON- CONSER VATIVE	interferon	Human Gene SWISSPROT- ID:Q16666 GAMMA-INTERFERON- INDUCIBLE PROTEIN IFI-16 (INTERFERON-INDUCIBLE MYELOID DIFFERENTIATION TRANSCRIPTIONAL ACTIVATOR) - HOMO SAPIENS (HUMAN), 729 aa.   pcds:SPTRMBL-ID:Q16666 IFI16=INTERFERON-INDUCIBLE MYELOID DIFFERENTIATION TRANSCRIPTIONAL ACTIVATOR - HOMO SAPIENS (HUMAN), 729 aa (fragment).	0	1

527	cg43090990	1083	TGCTCCATCAAAA ATGAAGCAAGGC C/TGCCATGTTA CCGACACCCGGA AAA	C	T	Pro	Leu (756)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1.-) (NPKC- THETA) - HOMO SAPIENS (HUMAN), 706 aa.	0	10
528	cg43969763	2663	CAAAAGCAAGAAA GTTCTTTGAGAA G/TJTGCCAGATG GCACCTTGGAACTT AA	G	T	Lys	Asn (757)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:Q13627 SERINE/THREONINE- SPECIFIC PROTEIN KINASE MINIBRAIN HOMOLOG (EC 2.7.1.-) (HP86) (DYRK) - HOMO SAPIENS (HUMAN), 763 aa.	0	21 (21q22.1 )
529	cg43932396	1226	AGTCCACCCGCG CCTCAGGCCCGTG C/C/TJGCTGGCCG AGTAGGAGAACT GGGGG	C	T	Gly	Ser (758)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:P31749 RAC-ALPHA SERINE/THREONINE KINASE (EC 2.7.1.-) (RAC-PK-ALPHA) (PROTEIN KINASE B) (PKB) (C-AKT) - HOMO SAPIENS (HUMAN), 480 aa.	1.40E-262	14 (14q32.3 )
530	cg43917871	1429	GGCACTGAAGAA ATCCCTGACATCA [T/C]ATTGGCGCT GCTGACGGGCGT ACTG	T	C	Met	Val (759)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)
531	cg43917871	1621	GGGCTGACAAGG TGCTGATTTTCAC T/GJGTGGACAAAG CGTTCCTCCATCGCT TT	T	G	Ser	Arg (760)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)

532	cg43917871	1713	TTCAATGTTGTAT TTGTCAATATAGTT /CJCATAATAATCTT CTGTCCCCAGAAC	T	C	Asp	Gly (761)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)
533	cg43917871	2096	TGTAATAATCGAAT ATCATAGTCTGTIT /GJAACGCTCGTA CAATTGCTTGAAG T	T	G	Leu	Phe (762)	NON- CONSER VATIVE	kinase	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	2.00E-215	11 (20p13)
534	cg43322545	1107	TCGGCTAGGCAG CCTCCATCCTCAC [A/C]CCCCCTTATCA CATCCGCGTGGC ATG	A	C	Thr	Pro (763)	NON- CONSER VATIVE	kinaserecep tor	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa. pcis:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	0	19 (19q13.1 )



536	cg43958558	863	TCCGGGATAAGCT CCAGGTGCTCCA G/TGGTAGGCGC CTGGAGGTGCCT GTCC	G	T	Pro	His (765)	NON- CONSER VATIVE	laminin	Human Gene Homologous to SWISSPROT-ID:P17931 GALECTIN- 3 (GALACTOSE-SPECIFIC LECTIN 3) (MAC-2 ANTIGEN) (IGE-BINDING PROTEIN) (35 KD LECTIN) (CARBOHYDRATE BINDING PROTEIN 35) (CBP 35) (LAMININ- BINDING PROTEIN) (LECTIN L-29) (L-31) (GALACTOSIDE-BINDING PROTEIN) (GALBP) - HOMO SAPIENS (HUMAN), 249 aa, pcsl:SWISSPROT-ID:P17931 GALECTIN-3 (GALACTOSE- SPECIFIC LECTIN 3) (MAC-2 ANTIGEN) (IGE-BINDING PROTEIN) (35 KD LECTIN) (CARBOHYDRATE BINDING PROTEIN 35) (CBP 35) (LAMININ-BINDING PROTEIN) (LECTIN L-29) (L-31) (GALACTOSIDE-BINDING PROTEIN) (GALBP) - HOMO SAPIENS (HUMAN), 249 aa.	3.90E-139	14 (14q21)
537	cg43966144	718	AAGCTTGTCATGC CTCACAGCAGTG C/AJGCACAAGACT GCCCAGCCCAAT GGA	C	A	Ala	Glu (766)	NON- CONSER VATIVE	MHC	Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	9.10E-147	6 (6p21.3)

538	cg43966144	823	ACTTACACCTGTG TGGTAGAGCACAC T/CJTGGGGCTCCT GAGCCCATCCTTC GG	T	C	Ile	Thr (767)	NON- CONSER VATIVE	MHC	Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	9.10E-147	6 (6p21.3)
539	cg42686658	907	GGCCTGGTGGGC TTCCTCGTGGGCA [C/T]CGTCTCATC ATCATGGGCACAT AT	C	T	Thr	Ile (768)	NON- CONSER VATIVE	MHC	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	3.70E-134	6 (6p21.3)
540	cg38337333	1044	CTGAGCCCGAG CGTTGTCTCCTGC [C/G]CATGAGCAC CACAGTCAGGCC TTGA	C	G	Pro	Ala (769)	NON- CONSER VATIVE	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
541	cg38337333	424	AGCCCGGCGGG CCCCACGGTTCG C/A/G]CAGGAGAG AACGTGACCTTGT CCTG	A	G	Thr	Ala (770)	NON- CONSER VATIVE	MHC	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	1.80E-113	19
542	cg42481172	340	CCGGCTGTGCTC AGGGGTGTGGGG T/A/G]CGGATACA GAGGAGCGGCTG GTGGA	A	G	Thr	Ala (771)	NON- CONSER VATIVE	misc_chann el	Human Gene Similar to SPTREMBL- ID:P91197 SIMILAR TO LIGAND- GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	2.30E-71	1



548	cg42732993	809	GGCTATAATCACA ATGGGGAATGGT G/TGTGAAGCCCAA ACCAAAAAATGGCC AA	G	T	Cys	Phe (777)	NON- CONSER VATIVE	oncogene	Human Gene Homologous to SPTREMBL-ID:Q13692 BCR/ABL FUSION PROTEIN - HOMO SAPIENS (HUMAN), 284 aa (fragment).	6.00E-150	
549	cg42904626	155	ATATAAACTTGTG GTAGTTGGAGCTT G/TGTGGCGTAG GCAAGAGTGCCTT GAC	G	T	Gly	Cys (778)	NON- CONSER VATIVE	oncogene	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K- RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	1.10E-97	12
550	cg42904626	304	TGGATATTCTCGA CACAGCAGGTCA A/CJGAGGAGTACA GTGCAATGAGGG ACC	A	C	Gln	His (779)	NON- CONSER VATIVE	oncogene	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K- RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	1.10E-97	12
551	cg42691989	706	CTGTTCCAGGATCT CCTCATTTCTGACJA /TJGTTCTCCTGAT GTCCAAATTGGTT G	A	T	Cys	Ser (780)	NON- CONSER VATIVE	peroxidase	Human Gene Homologous to SWISSPROT-ID:P18283 GLUTATHIONE PEROXIDASE- GASTROINTESTINAL (EC 1.11.1.9) (GSHPX-GI) (GLUTATHIONE PEROXIDASE-RELATED PROTEIN 2) (GPRP) - HOMO SAPIENS (HUMAN), 190 aa.	8.90E-101	14 (14q24.1 )



552	cg43917453	4096	AGGTCCTCGCGG AGCTGGGTCCGG G[A/G]CCCCGGAG GGTAGGTCAGCG CAGAC	A	G	Ser	Pro (781)	NON- CONSER VATIVE	phosphate	Human Gene TREMBLNEW- ID:G2262075 IAR/RECEPTOR-LIKE PROTEIN-TYROSINE PHOSPHATASE PRECURSOR - HOMO SAPIENS (HUMAN), 1015 aa.	0	7
553	cg43947363	368	CTGGCGCACTACT CGGACCTGCTCC[ C/T]CCTGGCGGG CCTGGGGCTGAT TGAG	C	T	Gly	Glu (782)	NON- CONSER VATIVE	phosphate	Human Gene SWISSPROT- ID:P23469 PROTEIN-TYROSINE PHOSPHATASE EPSILON PRECURSOR (EC 3.1.3.48) (R-PTP- EPSILON) - HOMO SAPIENS (HUMAN), 700 aa.	0	
554	cg43928335	3187	GCACAAGGAACG GAATTGCTGTCTG [A/G]TTTCTGCTTT AACAGCATTTGAT GC	A	G	Ile	Thr (783)	NON- CONSER VATIVE	phosphate	Human Gene SWISSPROT- ID:P54613 PROTEIN PHOSPHATASE PP2A, 65 KD REGULATORY SUBUNIT, BETA ISOFORM (PROTEIN PHOSPHATASE PP2A SUBUNIT A, BETA ISOFORM) (P65-BETA) - SUS SCROFA (PIG), 602 aa (fragment).	3.20E-302	11 (11q22)
555	cg43996195	1330	CTTCGGGGAAAG TTGGGGATTTCAC [C/T]GTAGTCAAAG ATCTGGGCCTGA GTT	C	T	Gly	Ser (784)	NON- CONSER VATIVE	phosphorylation	Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.	2.40E-155	

556	cg44022214	3340	AGGTCCTCCTCGA ATTGGGATGGCC[ A/G]AGGTGCATCA TCATCATCCCAGA GG	A	G	Trp	Arg (785)	NON- CONSER VATIVE	polymerase	Human Gene SWISSNEW- ID:P28340 DNA POLYMERASE DELTA CATALYTIC CHAIN (EC 2.7.7.7) - HOMO SAPIENS (HUMAN), 1107 aa.jpcls:SWISSPROT-ID:P28340 DNA POLYMERASE DELTA CATALYTIC CHAIN (EC 2.7.7.7) - HOMO SAPIENS (HUMAN), 1107 aa.	0	19 (19q13.3 )
557	cg43958858	1593	CTCAGACCATGTC CTTCGGATGCAC[ C/G]GTTACAGAGC ACCTGGGGAGCA GGA	C	G	Arg	Gly (786)	NON- CONSER VATIVE	polymerase	Human Gene SWISSNEW- ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN P1) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa.jpcls:SWISSPROT-ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN P1) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa.	0	6 (6p12)

558	cg42534568	641	CGCTTTGAGACG CAGCTGGGCACC C[AT]GGCGCAGT TCCCAACACACT CCTG	A	T	Gln	Leu (787)	NON- CONSER VATIVE	potassium_ channel	Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.5 (HK2) (HPCN1) - HOMO SAPIENS (HUMAN), 613 aa.	0	12 (12p13)
559	cg42534568	868	GGGGACGAGGC CATGGAGCGCTT C[C/G]GCGAGGAT GAGGCTTCATTA AAGA	C	G	Arg	Gly (788)	NON- CONSER VATIVE	potassium_ channel	Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.5 (HK2) (HPCN1) - HOMO SAPIENS (HUMAN), 613 aa.	0	12 (12p13)
560	cg42534568	910	CATTAAAGAAGAG GAGAAGCCCCTG[ C/G]CCCCGCAACG AGTCCAGCGCC AGGT	C	G	Pro	Ala (789)	NON- CONSER VATIVE	potassium_ channel	Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.5 (HK2) (HPCN1) - HOMO SAPIENS (HUMAN), 613 aa.	0	12 (12p13)
561	cg43154190	898	TGAGGGGATGC TCATTTGATGAA[ G/C]ATGAAAGGTG GACCAACAATTTC AG	G	C	Asp	His (790)	NON- CONSER VATIVE	protease	Human Gene Similar to SWISSPROT-ID:P50280 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE- 7) (MMP-7) (MATRIN) - RATTUS NORVEGICUS (RAT), 267 aa.	2.40E-59	11 (11q22)

562	cg43154190	923	GATGAAAGGTGG ACCAACAATTTCAT G/CJAGAGTACAAC TTACATCGTGTG CG	G	C	Arg	Thr (791)	NON- CONSER VATIVE	protease	Human Gene Similar to SWISSPROT-ID:P50280 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE- 7) (MMP-7) (MATRIN) - RATTUS NORVEGICUS (RAT), 267 aa.	2.40E-59	11 (11q22)
563	cg43927549	694	ATTCTACGATTCC GGTTTGCTCCAGI G/TJGTAACTAGC GCTCCTTTCCGTA AC	G	T	Gly	Cys (792)	NON- CONSER VATIVE	reductase	Human Gene Homologous to SWISSPROT-ID:P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT-DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.	1.60E-124	6 (6pter)
564	cg43325541	1081	CGCTGCCTTCTCC CGAAAGGTCTGCI C/TJCCCTTCACGCG TTCGGCTTCCCGC AG	C	T	Gly	Glu (793)	NON- CONSER VATIVE	synthase	Human Gene TREMBLNEW- ID:G2725625 ACETOLACTATE SYNTHASE - HOMO SAPIENS (HUMAN), 632 aa.	0	19

565	cg43064068	1474	GTGAAGGCATTG TGGTCCCTGGCCTT C/TTCAGTTCCTG TCCCATGACCCAG AA	C	T	Ser	Leu (794)	NON- CONSER VATIVE	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.lpcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.40E-65	
566	cg43064068	1617	GACTGTACACAGG GAAAATTCAACGA [G/A]CCCAAGCTTC GAGACAAGGAGT GGAA	G	A	Ala	Thr (795)	NON- CONSER VATIVE	synthase	Human Gene Similar to SWISSNEW- ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.lpcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE--COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL- COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.	7.40E-65	

567	cg36988276	1119	GCAAGAAGTTGAT TATATGACTCAG[A /G]CTAGGGGTCA GAGATCCTCTCTG GC	A	G	Thr	Ala (796)	NON- CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P23945 FOLLICLE STIMULATING HORMONE RECEPTOR PRECURSOR (FSH-R) (FOLLITROPIN RECEPTOR) - HOMO SAPIENS (HUMAN), 695 aa.	0	2 (2p21)
568	cg36988276	535	AAGGCCAACAAC CTGCTCTACATCA[ A/C]CCCTGAGGC CTTCCAGAACCTT CCC	A	C	Asn	Thr (797)	NON- CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P23945 FOLLICLE STIMULATING HORMONE RECEPTOR PRECURSOR (FSH-R) (FOLLITROPIN RECEPTOR) - HOMO SAPIENS (HUMAN), 695 aa.	0	2 (2p21)
569	cg32296848	1475	GAATGTCTTGAGA ATCCAGTGTCTC[ C/T]GCAGAAAGCA GTC TTCCAAACAT GC	C	T	Arg	Cys (798)	NON- CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P35348 ALPHA-1A ADRENERGIC RECEPTOR (ALPHA 1A-ADRENOCEPTOR) (ALPHA-1C ADRENERGIC RECEPTOR) - HOMO SAPIENS (HUMAN), 466 aa.	1.60E-252	8 (8p21)
570	cg2524739	1590	TCCTCTCTGGAGAA GATCCAACCCAT[ C/G]ACACAAAACG GTCAGCACCCAA CCT	C	G	Ile	Met (799)	NON- CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P21728 D(1A) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 446 aa.	8.30E-240	5 (5q35.1)

571	cg2320320	394	AGTGTCTGGATGA TCITTTGGGTCAJC /TTTGCATCCGTTT TCACAAATGGGCT T	C	T	Thr	Ile (800)	NON- CONSER VATIVE	tm7	Human Gene SWISSPROT- ID:P04001 GREEN-SENSITIVE OPSIN (GREEN CONE PHOTORECEPTOR PIGMENT) - HOMO SAPIENS (HUMAN), 364 aa.	8.50E-199	
572	cg43264978	519	CATCTTCTCCATC AACCTCTTCAGC[ A/G]GCATTTTCTT CCTCACGTGCATG AG	A	G	Ser	Gly (801)	NON- CONSER VATIVE	tm7	Human Gene TREMBLNEW- ID:G2736282 G PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 362 aa.	1.40E-196	
573	cg3003708	285	TCITTTGTGGACA TCITGCTTCTCCTTT /C]CACCCACCGTCC CCAAGATGCTGG CC	T	C	Phe	Ser (802)	NON- CONSER VATIVE	tm7	Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	2.50E-160	
574	cg38841806	68	GGCCCTGAGAGC AACACCACGGGC A/T]C]ACAGCCTT CTCCATGCCCCAG CTGG	T	C	Ile	Thr (803)	NON- CONSER VATIVE	tm7	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	2.10E-67	
575	cg43336100	688	TGGAAGCGTGCA TCCAGTGAGACCA [A/T]TGAGGCTTGA GTCTTTTAGTGCC TG	A	T	Met	Leu (804)	NON- CONSER VATIVE	tnf	Human Gene SWISSPROT- ID:P26022 PENTAXIN-RELATED PROTEIN PTX3 PRECURSOR (TUMOR NECROSIS FACTOR- INDUCIBLE PROTEIN TSG-14) - HOMO SAPIENS (HUMAN), 381 aa.	2.20E-207	3 (3q25)

576	cg43335562	234	GAGCGCGGGGA GCCAGGCCTGGG CT/C]CCGGGTCC CCAAGACCCCTGT GCTC	T	C	Leu	Pro (805)	NON- CONSER VATIVE	tnfreceptor	Human Gene Similar to TREMBLNEW-ID:G2653845 TNF RECEPTOR-RELATED RECEPTOR FOR TRAIL - HOMO SAPIENS (HUMAN), 386 aa.	2.30E-55	8
577	cg43140548	2857	ACTCGCACGTGG ATCCTGAGGCTGT [A/G]AGAGGTAAG GAAGGCTTTGCCA CAG	A	G	Tyr	His (806)	NON- CONSER VATIVE	transcriptfac tor	Human Gene SPTREMBL- ID:Q14872 METAL-REGULATORY TRANSCRIPTION FACTOR - HOMO SAPIENS (HUMAN), 753 aa.	0	1
578	cg43011561	1285	CATTGACAGCGA GGCCTCCTCAGC C[C/]TCTTCATGG CGAAGAAGAAGA CGCC	C	T	Leu	Phe (807)	NON- CONSER VATIVE	transcriptfac tor	Human Gene SWISSPROT- ID:P35269 TRANSCRIPTION INITIATION FACTOR IIF, ALPHA SUBUNIT (TFIIF-ALPHA) (TRANSCRIPTION INITIATION FACTOR RAP74) - HOMO SAPIENS (HUMAN), 517 aa.	4.30E-275	19 (19p13.3 )
579	cg43998970	1346	TGACAGAGCTGTA CCGTGACATTTTC /G]CAGCACCTTCG GGATGAATCAGG CA	C	G	Phe	Leu (808)	NON- CONSER VATIVE	transcriptfac tor	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	1.70E-177	12



580	cg2537639	464	CACTACTATGTCT TCACCGACCAAGC C/TGGCCGCGGT GCCCGCGGTGAC GCTG	T	Pro	Leu (809)	NON- CONSER VATIVE	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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582 583 584 585 586 587 588 589 590 591 592 593 594 595 596 597 598 599 600 601 602 603 604 605 606 607 608 609 610 611 612 613 614 615 616 617 618 619 620 621 622 623 624 625 626 627 628 629 630 631 632 633 634 635 636 637 638 639 640 641 642 643 644 645 646 647 648 649 650 651 652 653 654 655 656 657 658 659 660 661 662 663 664 665 666 667 668 669 670 671 672 673 674 675 676 677 678 679 680 681 682 683 684 685 686 687 688 689 690 691 692 693 694 695 696 697 698 699 700 701 702 703 704 705 706 707 708 709 710 711 712 713 714 715 716 717 718 719 720 721 722 723 724 725 726 727 728 729 730 731 732 733 734 735 736 737 738 739 740 741 742 743 744 745 746 747 748 749 750 751 752 753 754 755 756 757 758 759 760 761 762 763 764 765 766 767 768 769 770 771 772 773 774 775 776 777 778 779 780 781 782 783 784 785 786 787 788 789 790 791 792 793 794 795 796 797 798 799 800 801 802 803 804 805 806 807 808 809 810 811 812 813 814 815 816 817 818 819 820 821 822 823 824 825 826 827 828 829 830 831 832 833 834 835 836 837 838 839 840 841 842 843 844 845 846 847 848 849 850 851 852 853 854 855 856 857 858 859 860 861 862 863 864 865 866 867 868 869 870 871 872 873 874 875 876 877 878 879 880 881 882 883 884 885 886 887 888 889 890 891 892 893 894 895 896 897 898 899 900 901 902 903 904 905 906 907 908 909 910 911 912 913 914 915 916 917 918 919 920 921 922 923 924 925 926 927 928 929 930 931 932 933 934 935 936 937 938 939 940 941 942 943 944 945 946 947 948 949 950 951 952 953 954 955 956 957 958 959 960 961 962 963 964 965 966 967 968 969 970 971 972 973 974 975 976 977 978 979 980 981 982 983 984 985 986 987 988 989 990 991 992 993 994 995 996 997 998 999 1000

582	cg2537639	643	GGTGTGCGTGGA CGTGGACATGGA GTTATCCGCGAC CACGTGGGCGTG GAGAT	T	A	Phe	Ile (811)	NON- CONSER VATIVE	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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583 cg2537639 700 TCCGCTGTTCCG CACCCTGCACCC C[G/A]GCTTCTAC GGAAGCAGCCGG GAGGC

583	cg2537639	700	G	A	Gly	Ser (812)	NON- CONSER VATIVE	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYLTRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
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585	cg2537639	826	GTTCTTCGGGGG GTCGGTGCAAGA G[G/A]TGCAGCGG CTCACAGGGCC TGCCA	G	A	Val	Met (814)	NON- CONSER VATIVE	transferase	Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA- N- ACETYL GALACTOSAMINYL TRANS FERASE (EC 2.4.1.40) (HISTO- BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYL TRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	6.50E-192	9 (9q34)
586	cg42742340	3249	CACATCGTGGTG GAGCTGACCCAG G[C/A]TGACGCTTT GGGCTCCAGGTG GCGG	C	A	Ala	Asp (815)	NON- CONSER VATIVE	transport	Human Gene SWISSPROT- ID:Q04671 P PROTEIN (MELANOCYTE-SPECIFIC TRANSPORTER PROTEIN) - HOMO SAPIENS (HUMAN), 838 aa.	0	15
587	cg41653463	427	GTCTGAAAGATTT CCACAAGGACAT C[G]CTGAAGCCCT CACCAGGGAAGA GCC	C	G	Ile	Met (816)	NON- CONSER VATIVE	transport	Human Gene SWISSPROT- ID:P31641 SODIUM- AND CHLORIDE-DEPENDENT TAURINE TRANSPORTER - HOMO SAPIENS (HUMAN), 620 aa.	0	3 (3p25)

588	cg40351913	1165	CAAGTTCACCAAC AACTGCTACAGG G/CJACGCGATTGT CACCACCTCCATC AA	G	C	Asp	His (817)	NON- CONSER VATIVE	transport	Human Gene SWISSPROT- ID:Q01959 SODIUM-DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	0	5 (5p15.3)
589	cg40351913	1232	TCCTCCGGCTTCG TCGTCCTTCCTCT /CJCCCTGGGGTACA TGCCACAGAAAGC AC	T	C	Phe	Ser (818)	NON- CONSER VATIVE	transport	Human Gene SWISSPROT- ID:Q01959 SODIUM-DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	0	5 (5p15.3)
590	cg43955093	4776	CTGCGGGTAGCTG TCCCAGGCCCTCG G/C/GJCCGCGCCG CCTCGTCCATGTT GAGG	C	G	Ala	Pro (819)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	0	16
591	cg43055918	522	GCATAGGACATG GCGGGCTTGCCC C/C/GJCGCAGAGC TCTGGGGGCTAC TGCTA	C	G	Gly	Arg (820)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	0	17
592	cg43968854	4604	CAACCCCTAGAAG ACCTGGCTGGCTI T/G/GAAAGAGCTC TTCCAGACACCCAG TA	T	G	Leu	Trp (821)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSNEW- ACC:P46013 ANTIGEN KI-67 - Homo sapiens (Human), 3256 aa.	0	10 (10q25)

593	cg43070241	1841	CCATTGTTCAAGA CATCCTACGTTT /G/GAAATGCCCTGC AAGCAAAATTGTC C	T	G	Phe	Leu (822)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P55157 MICROSOMAL TRIGLYCERIDE TRANSFER PROTEIN, LARGE SUBUNIT PRECURSOR - Homo sapiens (Human), 894 aa.	0	4 (4q22)
594	cg43262121	2001	ACAAATTCAGAGAG GGAGACTGAGCA[ G/T]ACACCAGCAT TGATCATGGTGCC AA	G	T	Gln	His (823)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	0	
595	cg43262121	553	ATCAGGAAAGGT GTTGGATCACTGG [A/T]GCATCATGAC CAGTGAGGGAAGA AGT	A	T	Ser	Cys (824)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	0	
596	cg43262121	937	CCCCAAACAGGA AGTCCATGGGCC C[A/T]ACCCCTGACA GCAGCTTCTTAAC TTC	A	T	Asn	Tyr (825)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	0	
597	cg43262121	938	CCCCAAACAGGAA GTCCATGGGCC A[A/T]CCCTGACA GCAGCTTCTTAAC TTCC	A	T	Asn	Ile (826)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	0	



598	cg44024279	501	CTGGAAGAACCTTT GCCATGAGAAAG A/GJAATTTTGGAG AAGTACGGACATT CA	A	G	Glu	Gly (827)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P02771 ALPHA- FETOPROTEIN PRECURSOR (ALPHA-FETOGLOBULIN) (ALPHA- 1-FETOPROTEIN) - Homo sapiens (Human), 609 aa.	0	
599	cg44928804	1235	AATGATTAAACAC AACCTGAGACAC G/AJCGGATGAAAT GTTCTGGAACCCAC GT	G	A	Ala	Thr (828)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P21589 5'-NUCLEOTIDASE PRECURSOR (EC 3.1.3.5) (EC TO- NUCLEOTIDASE) (5'-NT) (CD73 ANTIGEN) - Homo sapiens (Human), 574 aa.	9.1e-313	6 (6q14)
600	cg43317253	367	GCCCCCAGGCAT GGCTAGCTCGTG TIG/TCCGTGCAG GTGAAGCTGGAG CTGGG	G	T	Ala	Ser (829)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSNEW- ACC:P42568 AF-9 PROTEIN - Homo sapiens (Human), 568 aa.	2.00E-301	9
601	cg41637661	223	CAGCTTCCATCC ATTTTATTATG/ AJACATACTGCT AGTGGAAGACCT A	G	A	Gly	Arg (830)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSNEW- ACC:O43913 ORIGIN RECOGNITION COMPLEX SUBUNIT 5 - Homo sapiens (Human), 435 aa.	6.10E-236	
602	cg42913861	3034	CAGGTGTCCTGC GAGCCACCCGGG G/AJCJTCCGGGTG GCGGGGGTGGCG GCGGC	A	C	Ser	Ala (831)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.	3.00E-227	2 (2cent)

603	cg43249389	526	AGAGGAGAGAGC CGCCCTCGAGCG G[A/G]GCAAGGCG ATTGAGAAAAACC TCAA	A	G	Ser	Gly (832)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P09471 GUANINE NUCLEOTIDE-BINDING PROTEIN (G(O), ALPHA SUBUNIT 1 - Homo sapiens (Human), 353 aa.	1.40E-188	15
604	cg43919239	335	GCCAGAGTTGCA GCATCAGGGCCA G[A/C]CTGAGCAG GAGACCCCCAGT CCCAT	A	C	Ser	Arg (833)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P14207 FOLATE RECEPTOR BETA PRECURSOR (FR-BETA) (FOLATE RECEPTOR 2) (FOLATE RECEPTOR, FETAL/PLACENTAL) (PLACENTAL FOLATE-BINDING PROTEIN) (FBP) - Homo sapiens (Human), 255 aa.	4.20E-150	
605	cg41642952	787	TAGGAATGACAGC AGTAGCAGTAAT A[G]GGAAGGCCA AAAATCCCCCTGG AGA	A	G	Arg	Gly (834)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P21583 STEM CELL FACTOR PRECURSOR (SCF) (MAST CELL GROWTH FACTOR) (MGF) (C-KIT LIGAND) - Homo sapiens (Human), 273 aa.	3.70E-142	12

606	cg43945147	221	TGTTCCCTGGAGCC TCAATGGTACAG G/CJGTGCTCGAG AAGGACAGTGTG ACTC	G	C	Arg	Ser (835)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:P08637 LOW AFFINITY IMMUNOGLOBULIN GAMMA FC RECEPTOR III-1 PRECURSOR (FC- GAMMA RIII) (FCRII) (IGG FC RECEPTOR III-1) (FC-GAMMA RIII-ALPHA) (CD16) (FCR-10) - Homo sapiens (Human), 254 aa.	1.60E-134	1
607	cg43926002	391	GGGCACAGAAAC ACAGCAGCGGGA G/C/SJAGCAACAC CAGCACTGCCAA CAGAT	C	S	Ser	??? (836)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Homologous to SWISSPROT-ACC:P50539 MAX INTERACTING PROTEIN 1 (MXI1 PROTEIN) - Homo sapiens (Human), 228 aa.	1.60E-116	10
608	cg43972311	1609	ATTGCCATTGTGG TAACCTCTGGGTC T/GJCATCATCTTC AGTGCCCCCAATTG TG	T	G	Glu	Ala (837)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD38008 GLYOXALASE-I (EC 4.4.1.5) - HOMO SAPIENS (HUMAN), 184 aa.	2.20E-98	6
609	cg42556108	521	GTGAAGCGGTGT ATGGGGACAGTG A/C/AJCCTCAACCA GGCCAGGGGCTC CTTT	C	A	Thr	Asn (838)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P49913 ANTIBACTERIAL PROTEIN FALL-39 PRECURSOR (FALL-39 PEPTIDE ANTIBIOTIC) (ANTIMICROBIAL PROTEIN CAP-18) (LL-37) - Homo sapiens (Human), 170 aa.	2.90E-87	3

610	cg36842490	487	AGTGACTTCAGTA AACTCTTGGGTC[ A/C]ACTTTCTGCC AAAAAGTACCTTG AG	A	C	Gln	Pro (839)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SWISSPROT-ACC:P01282 VASOACTIVE INTESTINAL PEPTIDE PRECURSOR (VIP) - Homo sapiens (Human), 170 aa.	2.30E-85	
611	cg43942549	1052	CGGTATAACGTCA AAAAATCCTGTTT[G /T]TCAGCCAAAGT TCAGAAATTGCCT C	G	T	Val	Phe (840)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:Q94218 CODED FOR BY C. ELEGANS CDNA CM10H5 - CAENORHABDITIS ELEGANS, 589 aa.	2.80E-73	4
612	cg42381630	283	AAGGCGCTATGTA CAGCCTCCTGAA[ A/G]TGATTGGGCC TATGCGGCCCGA GCA	A	G	Met	Val (841)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O76087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	5.90E-64	
613	cg42381630	505	TGAAGATGGTCCT GATGGCAGGAG[ A/G]TGGACCCGC CAAATCCAGAGGA GGT	A	G	Met	Val (842)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O76087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	5.90E-64	
614	cg3004395	260	ATTACTGAAGGGT GGAGAACAGAAAG[ G/C]GTCATGAAAA AATATCTGCTTCA TT	G	C	Gly	Arg (843)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G238693 T CELL RECEPTOR VARIABLE ALPHA CHAIN - HOMO SAPIENS (HUMAN), 143 aa (fragment).	1.00E-59	14 (14q11.2 )

615	cg43960645	733	CACITTCCTCTTC TCTTTGGATGCC[ A/T]CACCCCTCCTG TTGGGGGGCAGA TGG	A	T	Val	Glu (844)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to SWISSNEW- ACC:O76070 GAMMA-SYNUCLEIN (PERSYN) (BREAST CANCER- SPECIFIC GENE 1 PROTEIN) - Homo sapiens (Human), 127 aa.	1.20E-58	
616	cg2526759	289	GAAGACAAGGTG GTACAAAGCCCTC [T/A]ATCTCTGGTT GTCCACGAGGGA GAC	T	A	Leu	Gln (845)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	
617	cg2526759	342	TGTAACCTCTCAAT TGCAGTTATGAA[ G/A]TGACTAACTT TCGAAGCCCTACTA TG	G	A	Val	Met (846)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	
618	cg2526759	364	GAAGTGACTAACT TTCGAAGCCCTAC[ T/A]ATGGTACAAG CAGGAAAAGAAA GCT	T	A	Leu	Gln (847)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	
619	cg2526759	475	AGCATATTAGATA AGAAAAGAACTTTT /C]CAGCATCCTGA ACATCACAGCCAC C	T	C	Phe	Ser (848)	NON- CONSER VATIVE	UNCLASSI FIED	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	1.60E-54	

620	cg40310734	1067	TACAGAATATGTC GTCGGTGCCCCC[ gap/C]ACTTGGAG CTGGACCCCTGGG AGCGG	gap	C	Thr	His (849)	FRAMES HIFT	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
621	cg40310734	3285	GTCGGCTTCTTCA AGCGGAACCGGC[ gap/A]CACCCCTG GAAGAAGATGATG AAGA	gap	A	Pro	His (850)	FRAMES HIFT	cadherin	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	0	17 (17q21.3 2)
622	cg43956660	2521	GTCCATCACTTCA CTTCAGTTATTC[T/ gap]CCTAGGAGGT TGTATAGTCTTCT GA	T	gap	Arg	Glu (851)	FRAMES HIFT	cadherin	Human Gene SWISSNEW- ID:Q08722 LEUKOCYTE SURFACE ANTIGEN CD47 PRECURSOR (ANTIGENIC SURFACE DETERMINANT PROTEIN OA3) (INTEGRIN ASSOCIATED PROTEIN) (IAP) (MER6) - HOMO SAPIENS (HUMAN), 323 aa  pcis:SWISSPROT-ID:Q08722 LEUKOCYTE SURFACE ANTIGEN CD47 PRECURSOR (ANTIGENIC SURFACE DETERMINANT PROTEIN OA3) (INTEGRIN ASSOCIATED PROTEIN) (IAP) (MER6) - HOMO SAPIENS (HUMAN), 323 aa.	1.80E-157	

623	cg43970982	2429	CTCCAGGGATAGT TGGACAGAAGGG[ gap/GjAGACCCCTG GCTACCCAGGAC CAGCT	gap	G	Gly	Gly (852)	FRAMES HIFT	collagen	Human Gene SWISSPROT- ID:P12111 COLLAGEN ALPHA 3(VI) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 3176 aa.	0	2
624	cg42175288	1837	GCTATGGAGGCA AAATGGGAGGAA G[gap/GjAAACGAC TACAGAAATGATC AGCGC	gap	G	Arg	Arg (853)	FRAMES HIFT	dna_rna_bi nd	Human Gene SPTREMBL- ID:Q92804 PUTATIVE RNA BINDING PROTEIN RBP56 - HOMO SAPIENS (HUMAN), 592 aa.	0	17
625	cg42175288	263	CGGTTACTCCAGT TATGGACAAAAGT[ ap/CjTATTCACAGT CCTATGGTGGTTA TG	gap	C	Tyr	Leu (854)	FRAMES HIFT	dna_rna_bi nd	Human Gene SPTREMBL- ID:Q92804 PUTATIVE RNA BINDING PROTEIN RBP56 - HOMO SAPIENS (HUMAN), 592 aa.	0	17
626	cg41554010	584	GGCCGAGCAGCT GCGGCGCCAGCT G[gap/GjACCCCT ACGCACAGCGCA TGGAGA	gap	G	Thr	Asp (855)	FRAMES HIFT	eph	Human Gene SWISSNEW- ID:P06727 APOLOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.lpcis:SWISSPROT-ID:P06727 APOLOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	1.80E-203	11 (11q23)
627	cg43065549	1553	CAGACTTCCACAG AGTGCTGGATGA[ gap/AjCGCGGCCT GCCTTGCCCCAG GGTTA	gap	A	Thr	Asn (856)	FRAMES HIFT	glycoprotein	Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	0	15 (15q15)

628	cg41568631	999	TGACCCACGGGT GCTGGATGCCTG C[gap/C]TTATACAT CCTGGACCGGCG GGGGA	gap	C	Leu	Leu (857)	FRAMES HIFT	glycoprotein	Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	9.90E-70	14 (14q11.2 )
629	cg41637704	1220	GCGCCGCGAGAC AAGGGCAGCGGA C[gap/G]CGCCTGC GGACTTGAGGGA CAGTGA	gap	G	Pro	Arg (858)	FRAMES HIFT	homeobox	Human Gene SWISSPROT- ID:P50219 HOMEBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	1.20E-224	7
630	cg43933380	364	ATAAGTTACAATG CTTTTTTGTITIA/ gap)AAAAAAAAA AAAGTCTGTACTT TA	A	gap	Leu	End (859)	FRAMES HIFT	interferon	Human Gene SWISSPROT- ID:P15260 INTERFERON-GAMMA RECEPTOR ALPHA CHAIN PRECURSOR (CDW119) - HOMO SAPIENS (HUMAN), 489 aa.	1.40E-261	6
631	cg43072541	379	CTGTGGGGCTGG TTCTGTATCTGAT gap/CJATCATTCGA TTACGAAATAAAA CGT	gap	C	Ile	His (860)	FRAMES HIFT	kinase	Human Gene SPTREMBL- ID:Q15802 SERINE/THREONINE PROTEIN KINASE KRS-2 - HOMO SAPIENS (HUMAN), 487 aa.	9.60E-262	20



632	cg44032168	1536	GTCAGCCGCTAC CTCGACTGGATCC [gap/TTATGGGCAC ATCAGAGACAAG GAAGC	gap	T	His	Leu (861)	FRAMES HIFT	protease	Human Gene Similar to SWISSPROT-ID:P25155 COAGULATION FACTOR X PRECURSOR (EC 3.4.21.6) (STUART FACTOR) (VIRUS ACTIVATING PROTEASE) (VAP) - GALLUS GALLUS (CHICKEN), 475 aa.	2.40E-82	2 (2q13)
633	cg43931248	1317	CCGGGCAGAGCT GCGTCTGCTGAG G[gap/G]CTCAAGT TAAAAGTGGAGCA GCACG	gap	G	Leu	Ala (862)	FRAMES HIFT	tgf	Human Gene SWISSPROT- ID:P01137 TRANSFORMING GROWTH FACTOR BETA 1 PRECURSOR (TGF-BETA 1) - HOMO SAPIENS (HUMAN), 390 aa.	9.70E-214	19
634	cg43931248	1317	CCGGGCAGAGCT GCGTCTGCTGAG G[gap/G]CTCAAGT TAAAAGTGGAGCA GCACG	gap	G	Leu	Ala (863)	FRAMES HIFT	tgf	Human Gene SWISSPROT- ID:P01137 TRANSFORMING GROWTH FACTOR BETA 1 PRECURSOR (TGF-BETA 1) - HOMO SAPIENS (HUMAN), 390 aa.	9.70E-214	19
635	cg43272560	847	AATCTCCGCACTG CAGGCCAGGGC [gap/C]TGGCCAGC TACAGAGAGAGG TCACA	gap	C	Ala	Ala (864)	FRAMES HIFT	tgfreceptor	Human Gene SWISSPROT- ID:Q03167 TGF-BETA RECEPTOR TYPE III PRECURSOR (TGFR-3) (BETAGLYCAN) - HOMO SAPIENS (HUMAN), 849 aa.	0	1 (1p33)

636	cg43266471	1067	CCAGGATCCATT TGAGGATTATGG[ gap/T]GTGCTGGG ACACCATCAACTC CTCA	gap	T	Gly	Gly (865)	FRAMES HIFT	tm7	Human Gene SWISSPROT- ID:P32241 VASOACTIVE INTESTINAL POLYPEPTIDE RECEPTOR 1 PRECURSOR (VIP-R- 1) (PITUITARY ADENYLATE CYCLASE ACTIVATING POLYPEPTIDE TYPE II RECEPTOR) (PACAP-R-2) - HOMO SAPIENS (HUMAN), 457 aa.	5.20E-254	3
637	cg43995237	625	CAAATCCCCCGT TTCATCATCTTG[ ap/G]ACATGCTAA AATGAAATTACGC AGT	gap	G	Gln	Pro (866)	FRAMES HIFT	transferase	Human Gene SWISSPROT- ID:P53611 GERANYLGERANYL TRANSFERASE TYPE II BETA SUBUNIT (EC 2.5.1.-) (RAB GERANYLGERANYLTRANSFERAS E BETA SUBUNIT) (RAB GERANYL- GERANYLTRANSFERASE BETA SUBUNIT) (RAB GG TRANSFERASE) (RAB GGTASE) - HOMO SAPIENS (HUMAN), 331 aa.	1.40E-182	1

638	cg43995237	638	TTCTTCATCTTGA CATGCTAAATGCG ap/GJAAATTACGC AGTTTCTCTCTAT CAA	gap	G	Leu	Phe (867)	FRAMES HIFT	transferase	Human Gene SWISSPROT- ID:P53611 GERANYLGERANYL TRANSFERASE TYPE II BETA SUBUNIT (EC 2.5.1.-) (RAB GERANYLGERANYLTRANSFERAS E BETA SUBUNIT) (RAB GERANYL- GERANYLTRANSFERASE BETA SUBUNIT) (RAB GG TRANSFERASE) (RAB GGTASE) - HOMO SAPIENS (HUMAN), 331 aa.	1.40E-182	1
639	cg43254094	267	CCGCCTCTGCTG CTGCTGCTGCTG C[gap/G]GCGTCCC GCCCAGCCGCAG CTTCCC	gap	G	Arg	Arg (868)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P78539 SUSHI REPEAT- CONTAINING PROTEIN SRPX PRECURSOR - Homo sapiens (Human), 464 aa.	6.40E-257	X
640	cg44034555	665	ATCCAGGCTGAG CTGGATCATCTGA [G/gap]GGCCTCCA GCCACCCCGTTTTC CCTT	G	gap	Pro	Leu (869)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q13228 SELENIUM-BINDING PROTEIN 1 - Homo sapiens (Human), 472 aa.	3.80E-252	1
641	cg44034555	667	CCAGGCTGAGCT GGATCATCTGAG G[G/gap]CCTCCAG CCACCCGTTTTC CTTGA	G	gap	Gly	Gly (870)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSNEW- ACC:Q13228 SELENIUM-BINDING PROTEIN 1 - Homo sapiens (Human), 472 aa.	3.80E-252	1

642	cg39711096	882	AGCGAGTCCTCC GGGAGGCCACACA G[gap/G]TTACTGC CTCCAGCTGCAG CAGTGA	gap	G	Val	Gly (871)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P18428 LIPOPOLYSACCHARIDE-BINDING PROTEIN PRECURSOR (LBP) - Homo sapiens (Human), 481 aa.	1.00E-251	
643	cg44128902	379	CGTCCAGAGGA GCATATCTGCTGA [gap/C]TGATGACC TGCAAGAGTCATC CAGA	gap	C	Asp	Asp (872)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P18615 RD PROTEIN - Homo sapiens (Human), 380 aa.	1.00E-201	1 (1p36.2)
644	cg43946951	306	GGAACCTCAGCA CGTCGTCGGGGG A[C/gap]CCCAAGA TCACCGCGGCC TCTGGT	C	gap	Gly	Gly (873)	FRAMES HIFT	UNCLASSI FIED	Human Gene SWISSPROT- ACC:P09467 FRUCTOSE-1,6- BISPHOSPHATASE (EC 3.1.3.11) (D-FRUCTOSE-1,6- BISPHOSPHATE 1- PHOSPHOHYDROLASE) (FBPASE) - Homo sapiens (Human), 337 aa.	3.50E-178	9 (9q22.2)
645	cg43948890	195	ATTCGCCGGGGA GGGGGCCCTGTA A[G/gap]GGAACCC AGACAAATCCCATG AGACT	G	gap	Pro	Leu (874)	FRAMES HIFT	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:Q15182 SNRNP POLYPEPTIDE B - HOMO SAPIENS (HUMAN), 285 aa.	3.20E-147	20
646	cg43948890	197	TCCCGGGGGAGG GGGCCCTGTAAG G[G/gap]AAACCCAG ACAAATCCCATGAG ACTCC	G	gap	Phe	Phe (875)	FRAMES HIFT	UNCLASSI FIED	Human Gene Homologous to SPTREMBL-ACC:Q15182 SNRNP POLYPEPTIDE B - HOMO SAPIENS (HUMAN), 285 aa.	3.20E-147	20

647	cg43917524	713	GGGCCTGCTGC CCAGTGGAGGAG G[C/gap]TTCCTGCT GGTGTTCTAGGG GGCATC	C	gap	Ala	Pro (876)	FRAMES HIFT	UNCLASSI FIED	Human Gene Homologous to TREMBLNEW-ACC:AAD43025 PTD017 - HOMO SAPIENS (HUMAN), 258 aa.	3.20E-143	
648	cg43942004	373	CTCTCGGCACTG GTGACTGGCGAG A[gap/G]CCTGGAG CGGCTTCGGAGA GGGCTA	gap	G	Asp	Glu (877)	FRAMES HIFT	UNCLASSI FIED	Human Gene Homologous to SWISSNEW-ACC:Q99075 HEPARIN-BINDING EGF-LIKE GROWTH FACTOR PRECURSOR (HB-EGF) (HBEGF) (DIPHTERIA TOXIN RECEPTOR) (DT-R) - Homo sapiens (Human), 208 aa.	1.00E-107	5 (5q23)
649	cg43932428	681	TCGTGGCCAGGT CCTTCTGCCGTAAG [C/gap]CCCTTGCT CTGCCGACCTTG CTGGA	C	gap	Gly	Gly (878)	FRAMES HIFT	UNCLASSI FIED	Human Gene Similar to SPTREMBL- ACC:O60869 EDF-1 PROTEIN - HOMO SAPIENS (HUMAN), 148 aa.	2.50E-72	
650	cg44010855	450	GGTCCAAATGCAA GTGCTCCCGGAA[ G/gap]GGACCCAA GATCCGCTACAG CGACG	G	gap	Gly	Asp (879)	FRAMES HIFT	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD38944 NJAC PROTEIN - HOMO SAPIENS (HUMAN), 99 aa.	5.80E-50	5
651	cg44010855	452	TCCAAATGCAAGT GCTCCCGGAAGG[ G/gap]ACCCCAAGA TCCGCTACAGCG ACGTG	G	gap	Gly	Asp (880)	FRAMES HIFT	UNCLASSI FIED	Human Gene Similar to TREMBLNEW-ACC:AAD38944 NJAC PROTEIN - HOMO SAPIENS (HUMAN), 99 aa.	5.80E-50	5

# SEQUENCE LISTING

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Leach, Martin D.

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51

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51

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51

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51

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51

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51

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51

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51

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51

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51

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51

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51

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51

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51

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TAGCAGCCAG GTGACATGGC CAGGCTACCT TCCTGTACAG GCACTGTGGG C

51

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51

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CAGTGCATCT GGGAAGATTT CTACCCGACC AACAGTTCCT TCAGCTTCCA T

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51

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ACCGCACCCCT TTCCACCGGT GGGGGGCCCA GTGAAGTTTA ACAAAC TGCT G

51

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CATACCACGT TCACTGCAAG GGGGGGAACG TGTGGGTTC TCTATTCAAG A

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51

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51

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51

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51

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51

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51

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51

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51

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ACAAGACAGA AGCTGAAGTG CATCCCAAAG GTGCTCAGAG AGCCGGCCCCG C

51

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TCTTCCTTCT CCAGCCGGCA GGCCCGCGCC GCTTAGGAGG GAGAGCCCAC C

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51

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GTCCTTCGCT CCTGAGCACC AGGTTCAGTC TCCAGGAAGG GATTTGGTGA A

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CAAAAAAATT TATAAACTAA TTTTGGTACG TATGAATGAT ATCTTTGACC T

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CTGCCCTGCC ACCTGTCTGT CTGTCTCCAA AGAAGTTCTG GTATGAACTT G

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51

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51

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47

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51

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51

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51

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51

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51

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51

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51

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51

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ATTGTAGTAC AAATGACTCA CTGCTATAAA GCAGTTTTTC TACTTTTAAA G 51

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51

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51

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CCCACGCCTG CCAGGAGCAA GCCGAAGAGC CAGCCGGCCG GCGCACTCCG A

51

<210> 366

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<223> Accession number cg43928515

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51

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<223> Accession number cg43955093

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51

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51

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51

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51

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<223> Accession number cg44014448

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CTGCTCCCAA CTTCGCCAGC CTCCAGTGTA CAACTTCCGC GTGTAGTGGG C

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<223> Accession number cg43973129

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<223> Accession number cg44034555

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<212> DNA

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<222> (7)...(0)  
<223> cSNP translation

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Arg Leu His Arg Leu Arg Gly Glu Gln Met Ala Ser Tyr Phe  
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<210> 654  
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<223> cSNP translation

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Gly Ala Pro Leu Tyr Met Asp Ser Arg Ala Asp Arg Lys Leu  
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Gln Ala Gly Thr Thr Leu Asp Leu Asp Leu Gly Gly Lys His  
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Lys Cys Glu Cys Ser Arg Ala Tyr Gln Met Asp Leu Ala Thr  
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Asp Val Arg Gly Asn Leu Lys Gly Asn Thr Glu Gly Leu Gln  
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Pro Phe Gly Val Ile Val Arg Arg Gln Leu Asp Gly Arg Val  
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Trp Gly Lys Glu Pro Lys Leu Glu Ser Ala Trp Met Asn Gly  
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Gly Ala Met Leu Leu Asn Ile Ser Gly His Val Lys Glu Ser  
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Val Val Ile Pro Ser Asp Phe Phe Gln Ile Val Gly Gly Ser  
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Lys Arg Lys Leu Phe Ile Arg Ser Met Gly Glu Gly Thr Ile  
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Glu Gln Ala Arg Gln Gly Leu Lys Gly Leu Glu Glu Thr Val  
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<210> 668  
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Cys End Ala Gln Ser Val Ile Ser Cys Pro End Ala Pro Gln  
1 5 10

<210> 669  
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Ser Phe Leu Ile Ser Pro Leu Thr Pro Ala His Ala Gly Thr  
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Trp Cys Ser Lys Lys Lys Asp Ala Ala Val Met Asn Gln Glu  
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<210> 671

<211> 14

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<222> (7)...(0)

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<222> (7)...(0)

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Gly Ile Gln Asn Lys Glu Val Glu Val Arg Ile Phe His Cys  
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<222> (7)...(0)

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<400> 673

Val Asp Asn Ile Arg Ser Val Phe Gly Asn Ala Val Ser Arg  
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Leu Thr Ile Gln Leu Ile Glu Asn His Phe Val Asp Glu Tyr  
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<223> cSNP translation

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<400> 686

Asp Pro Leu Ile Tyr Ala Phe Arg Ser Gln Glu Met Arg Lys  
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1 5 10

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<400> 700  
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<210> 701  
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<400> 701  
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<400> 702  
Trp Leu Gly Phe Asn Lys Gln Arg Gly His Leu Gln Ile Ala  
1 5 10

<210> 703

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<400> 703  
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1 5 10

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<400> 704  
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<400> 705  
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<400> 706  
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<400> 707  
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1 5 10

<210> 708  
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<400> 708  
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1 5 10

<210> 709  
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<210> 710  
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<400> 710  
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<400> 711

Pro Val Met Gly Leu Met Ile Tyr Met Met Val Met Asp His  
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Ser Ser Gln Asp Pro Ala Ser Val Arg Glu Cys His Asp Pro  
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<210> 713

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<400> 713

Val Leu Leu Leu Leu Gly Ala Cys Ala Ala Pro Pro Ala Trp  
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<210> 714

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<400> 714

Ser Leu Pro Tyr Ala Val Ala Pro Leu Ser Leu Pro Arg Gly  
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<210> 715

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<400> 715  
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1 5 10

<210> 716  
<211> 14  
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<400> 716  
Thr Met Asp Cys Thr His Ser Leu Gly Asn Phe Ser Phe Ser  
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<210> 717  
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<400> 717  
Gly Thr Thr Glu Thr Gly Gly Gln Gly Lys Gly Thr Ser Lys  
1 5 10

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<400> 718  
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<210> 719  
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<400> 719

Leu Val Ser Tyr Cys Pro Arg Arg Leu Gln Gln Leu Leu Pro  
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<210> 720

<211> 14

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<210> 721

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<400> 721

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<400> 727  
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Ile Asn Ala Tyr Ile Ser His Leu Gly Phe Arg Phe  
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<210> 729  
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Leu His Ser Gly His Arg Gln Arg Pro Glu Phe Arg Pro Tyr  
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<210> 730  
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<210> 731  
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<400> 731

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<210> 732

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<210> 735

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<210> 737  
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<210> 738  
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1 5 10

<210> 739  
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<210> 740  
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1 5 10

<210> 741  
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1 5 10

<210> 742  
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1 5 10

<210> 743  
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<400> 743



Thr Glu Ile Thr Gly Ala Thr Thr Met Thr Ser Val Gly His  
1 5 10

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<210> 745  
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1 5 10

<210> 746  
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<210> 747  
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1 5 10

<210> 748  
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<210> 749  
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<220>  
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<210> 750  
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<220>  
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<400> 750  
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<210> 751  
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<220>  
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<400> 751  
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<210> 752  
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<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 752  
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1 5 10

<210> 753  
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<220>  
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<222> (7)...(0)  
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<400> 753  
Cys Leu Thr Glu Arg Gln Ser Lys Ile Trp Phe Gln Asn Arg  
1 5 10

<210> 754  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
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<400> 754  
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1 5 10

<210> 755  
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<222> (7)...(0)  
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<400> 755  
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1 5 10

<210> 756  
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<220>  
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<222> (7)...(0)  
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<400> 756  
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1 5 10

<210> 757  
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<400> 757  
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1 5 10

<210> 758  
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<220>  
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<223> cSNP translation

<400> 758  
Phe Ser Tyr Ser Ala Ser Ser Thr  
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<210> 759  
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<220>  
<221> VARIANT  
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<400> 759  
Pro Val Ser Ser Ala Asn Val Met Ser Gly Ile Ser Ser Val  
1 5 10

<210> 760

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<220>  
<221> VARIANT  
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<400> 760  
Trp Glu Arg Phe Val His Arg Glu Asn Gln His Leu Val Ser  
1 5 10

<210> 761  
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<222> (7)...(0)  
<223> cSNP translation

<400> 761  
Gly Thr Glu Asp Leu Tyr Gly Tyr Ile Asp Lys Tyr Asn Ile  
1 5 10

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Lys Gln Leu Tyr Gln Thr Phe Thr Asp Tyr Asp Ile Arg Phe  
1 5 10

<210> 763  
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Gly Ser Leu His Pro His Pro Pro Tyr His Ile Arg Val Ala  
1 5 10

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Leu Tyr Ser Arg Leu Gly Gly Gln Pro Val Tyr Leu Pro Thr  
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<210> 765  
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Ala Pro Pro Gly Ala Tyr His Gly Ala Pro Gly Ala Tyr Pro  
1 5 10

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Val Met Pro His Ser Ser Glu His Lys Thr Ala Gln Pro Asn  
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<210> 767  
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1 5 10

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<400> 768

Val Gly Phe Leu Val Gly Ile Val Leu Ile Ile Met Gly Thr  
1 5 10

<210> 769

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<400> 769

Gln Ser Val Val Ser Cys Ala End Ala Pro Gln Ser Gly Leu  
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<400> 770

Pro Gly Pro Thr Val Arg Ala Gly Glu Asn Val Thr Leu Ser  
1 5 10

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<400> 771

Cys Ser Gly Val Trp Gly Ala Asp Thr Glu Glu Arg Leu Val  
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<210> 773  
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Gly Phe His Tyr Gly Val Leu Ala Cys Glu Gly Cys Lys Gly  
1 5 10

<210> 775  
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Asp Gly Pro Glu Gln Ala His Arg Gln Arg Arg Arg Gln Arg  
1 5 10

<210> 776  
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<220>  
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Glu End Ala Met Ser Trp Phe Leu Phe Leu Ala His Arg Val  
1 5 10

<210> 777  
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<210> 778  
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<210> 779  
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<400> 779  
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1 5 10

<210> 780  
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<400> 780  
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1 5 10

<210> 781  
<211> 14  
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1 5 10

<210> 782  
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<400> 782  
Gln Pro Gln Ala Arg Gln Glu Glu Gln Val Arg Val Val Arg  
1 5 10

<210> 783  
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<400> 783  
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1 5 10

<210> 784  
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<223> cSNP translation

<400> 784

Ala Gln Ile Phe Asp Tyr Ser Glu Ile Pro Asn Phe Pro Arg  
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<210> 785

<211> 14

<212> PRT

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<400> 785

Asp Asp Asp Asp Ala Pro Arg Pro Ser Gln Phe Glu Glu Asp  
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<210> 786

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<400> 786

His Val Leu Arg Met His Gly Tyr Arg Ala Pro Gly Glu Gln  
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<210> 787

<211> 14

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<222> (7)...(0)

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<400> 787

Glu Thr Gln Leu Gly Thr Leu Ala Gln Phe Pro Asn Thr Leu  
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<210> 788

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<400> 788

Glu Ala Met Glu Arg Phe Gly Glu Asp Glu Gly Phe Ile Lys  
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<210> 789

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<400> 789

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1 5 10

<210> 790

<211> 14

<212> PRT

<213> Homo sapiens

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<400> 790

Asp Ala His Phe Asp Glu His Glu Arg Trp Thr Asn Asn Phe  
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<210> 791

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<212> PRT

<213> Homo sapiens

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<400> 791

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<210> 793  
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1 5 10

<210> 794  
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<210> 795  
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<400> 795  
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<210> 796  
<211> 14  
<212> PRT  
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<220>  
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<210> 797  
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1 5 10

<210> 798  
<211> 14  
<212> PRT  
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Leu Arg Ile Gln Cys Leu Cys Arg Lys Gln Ser Ser Lys His  
1 5 10

<210> 799  
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1 5 10

<210> 800  
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<400> 800

Trp Met Ile Phe Val Val Ile Ala Ser Val Phe Thr Asn Gly  
1 5 10

<210> 801  
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<212> PRT  
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<220>  
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<400> 801  
Ser Ile Asn Leu Phe Ser Gly Ile Phe Phe Leu Thr Cys Met  
1 5 10

<210> 802  
<211> 14  
<212> PRT  
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<400> 802  
Val Asp Ile Cys Phe Ser Ser Thr Thr Val Pro Lys Met Leu  
1 5 10

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<400> 803  
Glu Ser Asn Thr Thr Gly Thr Thr Ala Phe Ser Met Pro Ser  
1 5 10

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<400> 804  
Val His Pro Val Arg Pro Leu Arg Leu Glu Ser Phe Ser Ala

1 5 10

<210> 805  
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<400> 805  
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1 5 10

<210> 806  
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<400> 806  
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1 5 10

<210> 807  
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<400> 807  
Ser Glu Ala Ser Ser Ala Phe Phe Met Ala Lys Lys Lys Thr  
1 5 10

<210> 808  
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<400> 808  
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1 5 10



<210> 809  
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<400> 809  
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1 5 10

<210> 810  
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<400> 810  
Leu Ser Val Leu Glu Val Gly Ala Tyr Lys Arg Trp Gln Asp  
1 5 10

<210> 811  
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<400> 811  
Val Asp Val Asp Met Glu Ile Arg Asp His Val Gly Val Glu  
1 5 10

<210> 812  
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<400> 812  
Phe Gly Thr Leu His Pro Ser Phe Tyr Gly Ser Ser Arg Glu  
1 5 10

<210> 813  
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<400> 813  
Glu Gly Asp Phe Tyr Tyr Met Gly Gly Phe Phe Gly Gly Ser  
1 5 10

<210> 814  
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<400> 814  
Gly Gly Ser Val Gln Glu Met Gln Arg Leu Thr Arg Ala Cys  
1 5 10

<210> 815  
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<400> 815  
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1 5 10

<210> 816  
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<400> 816  
Lys Asp Phe His Lys Asp Met Leu Lys Pro Ser Pro Gly Lys  
1 5 10

<210> 817

<211> 14  
<212> PRT  
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<400> 817  
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1 5 10

<210> 818  
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<400> 818  
Gly Phe Val Val Phe Ser Ser Leu Gly Tyr Met Ala Gln Lys  
1 5 10

<210> 819  
<211> 14  
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<400> 819  
Met Asp Glu Ala Ala Arg Pro Glu Ala Trp Asp Ser Tyr Arg  
1 5 10

<210> 820  
<211> 14  
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<220>  
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<400> 820  
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1 5 10

<210> 821  
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<400> 821  
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1 5 10

<210> 822  
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<400> 822  
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1 5 10

<210> 823  
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<400> 823  
Ser Glu Arg Glu Thr Glu His Thr Pro Ala Leu Ile Met Val  
1 5 10

<210> 824  
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<212> PRT  
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<400> 824  
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1 5 10

<210> 825  
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<213> Homo sapiens

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<400> 825

Gln Glu Val His Gly Pro Tyr Pro Asp Ser Ser Phe Leu Thr  
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<210> 826

<211> 14

<212> PRT

<213> Homo sapiens

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<222> (7)...(0)

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<400> 826

Gln Glu Val His Gly Pro Ile Pro Asp Ser Ser Phe Leu Thr  
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<210> 827

<211> 14

<212> PRT

<213> Homo sapiens

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<222> (7)...(0)

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<400> 827

Glu Leu Cys His Glu Lys Gly Ile Leu Glu Lys Tyr Gly His  
1 5 10

<210> 828

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 828

Asn Asn Asn Leu Arg His Thr Asp Glu Met Phe Trp Asn His  
1 5 10

<210> 829

<211> 14

<212> PRT

<213> Homo sapiens

<220>  
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<223> cSNP translation

<400> 829  
Gly Met Ala Ser Ser Cys Ser Val Gln Val Lys Leu Glu Leu  
1 5 10

<210> 830  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
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<400> 830  
Pro Ser Ile Phe Ile Tyr Arg His Thr Ala Ser Gly Lys Thr  
1 5 10

<210> 831  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
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<400> 831  
Pro Pro Pro Pro Pro Gly Ala Pro Gly Gly Ser Gln Asp Thr  
1 5 10

<210> 832  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 832  
Arg Ala Ala Leu Glu Arg Gly Lys Ala Ile Glu Lys Asn Leu  
1 5 10

<210> 833  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 833  
Thr Gly Gly Leu Leu Leu Arg Leu Ala Leu Met Leu Gln Leu  
1 5 10

<210> 834  
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<220>  
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<400> 834  
Asp Ser Ser Ser Ser Asn Gly Lys Ala Lys Asn Pro Pro Gly  
1 5 10

<210> 835  
<211> 14  
<212> PRT  
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<220>  
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<400> 835  
Leu Glu Pro Gln Trp Tyr Ser Val Leu Glu Lys Asp Ser Val  
1 5 10

<210> 836  
<211> 14  
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<220>  
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<400> 836  
Gln Lys His Ser Ser Gly Xaa Ser Asn Thr Ser Thr Ala Asn  
1 5 10

<210> 837  
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<400> 837  
Trp Gly Thr Glu Asp Asp Ala Thr Gln Ser Tyr His Asn Gly  
1 5 10

<210> 838  
<211> 14  
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<220>  
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<400> 838  
Arg Cys Met Gly Thr Val Asn Leu Asn Gln Ala Arg Gly Ser  
1 5 10

<210> 839  
<211> 14  
<212> PRT  
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<400> 839  
Phe Ser Lys Leu Leu Gly Pro Leu Ser Ala Lys Lys Tyr Leu  
1 5 10

<210> 840  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 840  
Thr Ser Lys Ile Leu Phe Phe Ser Gln Gly Ser Glu Ile Ala  
1 5 10

<210> 841  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)

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<400> 841

Tyr Val Gln Pro Pro Glu Val Ile Gly Pro Met Arg Pro Glu  
1 5 10

<210> 842

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 842

Gly Pro Asp Gly Gln Glu Val Asp Pro Pro Asn Pro Glu Glu  
1 5 10

<210> 843

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 843

Lys Gly Gly Glu Gln Lys Arg His Glu Lys Ile Ser Ala Ser  
1 5 10

<210> 844

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 844

Ala Pro Gln Gln Glu Gly Glu Ala Ser Lys Glu Lys Glu Glu  
1 5 10

<210> 845

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 845

Lys Val Val Gln Ser Pro Gln Ser Leu Val Val His Glu Gly  
1 5 10

<210> 846

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 846

Leu Asn Cys Ser Tyr Glu Met Thr Asn Phe Arg Ser Leu Leu  
1 5 10

<210> 847

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 847

Thr Asn Phe Arg Ser Leu Gln Trp Tyr Lys Gln Glu Lys Lys  
1 5 10

<210> 848

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 848

Leu Asp Lys Lys Glu Leu Ser Ser Ile Leu Asn Ile Thr Ala  
1 5 10

<210> 849

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (8)...(0)

<223> cSNP translation

<400> 849  
Glu Tyr Val Val Gly Ala Pro His Leu Glu Leu Asp Pro Gly  
1 5 10

<210> 850  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 850  
Phe Phe Lys Arg Asn Arg His Thr Pro Gly Arg Arg End End  
1 5 10

<210> 851  
<211> 9  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 851  
Thr Ile Gln Pro Pro Arg Glu End Leu  
1 5

<210> 852  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 852  
Gly Ile Val Gly Gln Lys Gly Arg Pro Trp Leu Pro Arg Thr  
1 5 10

<210> 853  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 853  
Gly Gly Lys Met Gly Gly Arg Lys Arg Leu Gln Lys End Ser  
1 5 10

<210> 854  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 854  
Tyr Ser Ser Tyr Gly Gln Ser Leu Phe Thr Val Leu Trp Trp  
1 5 10

<210> 855  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 855  
Glu Gln Leu Arg Arg Gln Leu Asp Pro Leu Arg Thr Ala His  
1 5 10

<210> 856  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 856  
Ser Thr Glu Cys Trp Met Asn Ala Ala Cys Leu Ala Pro Gly  
1 5 10

<210> 857  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (9)...(0)  
<223> cSNP translation

<400> 857

His Gly Val Leu Asp Ala Cys Leu Ile His Pro Gly Pro Ala  
1 5 10

<210> 858  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 858  
Arg Asp Lys Gly Ser Gly Arg Ala Cys Gly Leu Glu Gly Gln  
1 5 10

<210> 859  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 859  
Thr Asp Phe Phe Phe End Thr Lys Lys Ala Leu End Leu  
1 5 10

<210> 860  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 860  
Gly Ala Gly Ser Val Ser Asp His His Ser Ile Thr Lys End  
1 5 10

<210> 861  
<211> 12  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 861  
Arg Tyr Leu Asp Trp Ile Leu Trp Ala His Gln Arg

1 5 10

<210> 862  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 862  
Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala  
1 5 10

<210> 863  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 863  
Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala  
1 5 10

<210> 864  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 864  
Pro His Cys Arg Pro Gly Ala Trp Pro Ala Thr Glu Arg Gly  
1 5 10

<210> 865  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 865  
Ile His Phe Glu Asp Tyr Gly Val Leu Gly His His Gln Leu  
1 5 10

<210> 866  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 866  
Asn Phe Ile Leu Ala Cys Pro Arg End Arg Asn Gly Gly Ile  
1 5 10

<210> 867  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 867  
Arg Glu Lys Leu Arg Asn Phe His Phe Ser Met Ser Arg End  
1 5 10

<210> 868  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 868  
Leu Leu Leu Leu Leu Leu Arg Arg Pro Ala Gln Pro Gln Leu  
1 5 10

<210> 869  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 869  
Lys Arg Val Ala Gly Gly Leu Arg End Ser Ser Ser Ala Trp  
1 5 10

<210> 870  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 870  
Gly Lys Arg Val Ala Gly Gly Leu Arg End Ser Ser Ser Ala  
1 5 10

<210> 871  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 871  
Ser Ser Gly Arg Pro Thr Gly Tyr Cys Leu Gln Leu Gln Gln  
1 5 10

<210> 872  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 872  
Gln Arg Ser Ile Ser Ala Asp End End Pro Ala Arg Val Ile  
1 5 10

<210> 873  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 873  
Arg Ala Pro Val Ile Leu Gly Pro Pro Thr Thr Cys Ser Ser  
1 5 10

<210> 874



<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 874  
Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro Gly  
1 5 10

<210> 875  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 875  
Leu Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro Pro  
1 5 10

<210> 876  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 876  
Pro Arg Thr Pro Ala Glu Pro Pro Pro Leu Gly Arg Gln Ala  
1 5 10

<210> 877  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 877  
Gly Thr Gly Asp Trp Arg Glu Pro Gly Ala Ala Ser Glu Arg  
1 5 10

<210> 878  
<211> 14

<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (9)...(0)  
<223> cSNP translation

<400> 878  
Gln Gly Arg Gln Ser Lys Gly Leu Arg Arg Arg Thr Trp Pro  
1 5 10

<210> 879  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 879  
Lys Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala  
1 5 10

<210> 880  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 880  
Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala Thr  
1 5 10

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